COVERING LETTER

To

The Dean,

IHEC-PGR, AIIMS BHOPAL

Respected sir,

Sub: Submission of thesis

I, **Dr.Priya Maheshwari**, who have joined the Department of Orthopaedics, AIIMS Bhopal as a Post Graduate in **July 2022** session with Registration number: **22002437M0P31010.**

I am hereb y submitting my thesis titled **"Spectrum of Patients with Congenital Limb Defects presenting at a Tertiary Health Care Hospital"** for your kind approval.

Thanking you

Yours faithufully,

Dr. Priya Maheshwari

Guide:

Dr. John A Santoshi,

Professor,

Department of Orthopaedics

AIIMS Bhopal

डॉ.जॉन ए संतोषी/Dr. John A. Santoshi
MS Orth, DNB Orth, F Hand Surg. /Reg.No.MP11493
प्राध्यापक/Professor
अस्थिरोग विभाग/Department of Orthopaedics
अखिल भारतीय आयुर्विज्ञान संस्थान, भोपाल-462020
All India Institute of Medical Sciences Bhopal-462020

Place: AIIMS Bhopal

Date: 15/10/24

All India Institute of Medical Sciences, Bhopal
Institutional Human Ethics Committee-Student Research

"SPECTRUM OF PATIENTS WITH CONGENITAL LIMB DEFECTS PRESENTING AT A TERTIARY HEALTH CARE HOSPITAL"

A thesis submitted to the faculty of

ALL INDIA INSTITUTE OF MEDICAL SCIENCES, BHOPAL

For the award of the degree MS in the subject of

Orthopaedics

By

Dr. Priya Maheshwari
(Reg No: 2200437M02P31010)

Under the guidance of

Dr. John A Santoshi,
MS Orthopaedics,
Professor,
Department of Orthopaedics,
AIIMS, Bhopal.

(October 2024)

ALL INDIA INSTITUTE OF MEDICAL SCIENCES, BHOPAL

UNDERTAKING

I. **Dr. Priya Maheshwari,** MS scholar registered in the batch **July 2022** with **Registration No: 2200437M02P31010** hereby declare that the data submitted for my MS thesis belongs to me and it is original. I also certify that the work was done by me to carry out this study.

The Institute shall not be responsible for any dispute or any conflict of interest that may arise out of this thesis at any stage.

Dr. Priya Maheshwari

Head of the Department:
Dr. Rehan Ul Haq,
Professor and Head
Department of Orthopaedics,
AIIMS, Bhopal.

Dr. REHAN UL HAQ
Reg.No.MP-2331, MS Ortho.,MNAMS
Professor & Head
Department of Orthopaedics
AIIMS, Bhopal (M.P.)-462020

Place: AIIMS, Bhopal.
Date: 15/10/24

ALL INDIA INSTITUTE OF MEDICAL SCIENCES, BHOPAL

UNDERTAKING

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Priya Maheshwari 15/10/24

Dr. Priya Maheshwari

Guide:

Dr. John A Santoshi,

Professor,

Department of Orthopaedics,

AIIMS, Bhopal.

Dr. John A. Santoshi
MS Orth, DNB Orth, F Hand Surg / Reg No.MP11493
प्राध्यापक / Professor
अस्थिरोग विभाग / Department of Orthopaedics
अखिल भारतीय आयुर्विज्ञान संस्थान भोपाल-462020
All India Institute of Medical Sciences Bhopal-462020

Place: AIIMS Bhopal

Date: 15/10/24

All India Institute of Medical Sciences, Bhopal
Institutional Human Ethics Committee-Student Research

ALL INDIA INSTITUTE OF MEDICAL SCIENCES, BHOPAL

CERTIFICATE

This is to certify that the thesis titled **"SPECTRUM OF PATIENTS WITH CONGENITAL LIMB DEFECTS PRESENTING AT A TERTIARY HEALTH CARE HOSPITAL"** is bonafide and genuine research carried out by Dr. Priya Maheshwari under our guidance and direct supervision for the award of MS.

Name of the Guide and Designation:

Dr. John A Santoshi,
Professor,
Department of Orthopaedics,
AIIMS, Bhopal.

डॉ. ए संतोषी / Dr. John A. Santoshi
MS Orth, DNB Orth, F Hand Surg / Reg No.MP11493
प्राध्यापक / Professor
अस्थिरोग विभाग / Department of Orthopaedics
अखिल भारतीय आयुर्विज्ञान संस्थान भोपाल-462020
All India Institute of Medical Sciences Bhopal-462020

Dr. Shikha Malik,
Professor,
Department of Pediatrics,
AIIMS, Bhopal

15.10.2024

डॉ. शिखा मलिक
रजि. न. M.P.-11453
प्राध्यापक एवं विभागाध्यक्ष
बाल्य एवं शिशु रोग विभाग
अखिल भारतीय आयुर्विज्ञान संस्थान
साकेत नगर, भोपाल-462020 (म.प्र.)

Names of co-guides and designation:

Dr. Prateek Behera,
Additional Professor,
Department of Orthopaedics,
AIIMS, Bhopal

डॉ. प्रतीक बेहेरा / Dr. PRATEEK BEHERA
Reg.No.16818/08 MS, DNB, MNAMS
अपर प्राध्यापक / Additional Professor
अस्थिरोग विभाग / Department of Orthopaedics
अखिल भारतीय आयुर्विज्ञान संस्थान भोपाल-462020
All India Institute of Medical Sciences Bhopal-462020

Dr. Manal M Khan,
Professor,
Department of Burns and Plastic Surgery
AIIMS, Bhopal

डॉ. मनाल एम. खान / Dr. MANAL M. KHAN
M.S.,M.Ch.(Plastic & Reconst. Surgery) Reg.No.25149
प्राध्यापक एवं विभागाध्यक्ष / Professor & HOD
प्लास्टिक सर्जरी विभाग/Deptt. of Burns & Plastic Surgery
अखिल भारतीय आयुर्विज्ञान संस्थान भोपाल-462020
All India Institute of Medical Sciences Bhopal-462020

Place: AIIMS Bhopal
Date: 15/10/24

ALL INDIA INSTITUTE OF MEDICAL SCIENCES, BHOPAL

Declaration

I hereby declare that the thesis **"SPECTRUM OF PATIENTS WITH CONGENITAL LIMB DEFECTS PRESENTING AT A TERTIARY HEALTH CARE HOSPITAL"** is a bonafide and original work carried out by me under the supervision and guidance of **Dr. John A Santoshi** Professor, Department of Orthopaedics and Co-guides **Dr. Prateek Behera,** Associate Professor, Department of Orthopaedics AIIMS, Bhopal. **Dr. Shikha Malik,** Professor, Department of Pediatrics, AIIMS, Bhopal. **Dr Manal M Khan,** Professor, Department of Burns and Plastic Surgery, AIIMS Bhopal. The thesis or any part thereof has not formed the basis for the award of any degree/fellowship or similar title to any candidate of any University to the best of my knowledge.

Name of the student: Priya Maheshwari
Dr. Priya Maheshwari

Place: AIIMS Bhopal
Date: 15/10/24

ALL INDIA INSTITUTE OF MEDICAL SCIENCES, BHOPAL

CERTIFICATE

This is to certify that the thesis entitled **"SPECTRUM OF PATIENTS WITH CONGENITAL LIMB DEFECTS PRESENTING AT A TERTIARY HEALTH CARE HOSPITAL"** is a bonafide and original work carried out by **Dr. Priya Maheshwari** under the supervision and guidance of **Dr. John A Santoshi** Professor, Department of Orthopaedics and Co-guides **Dr. Prateek Behera,** Additional Professor, Department of Orthopaedics AIIMS, Bhopal. **Dr. Shikha Malik,** Professor, Department of Pediatrics, AIIMS, Bhopal. **Dr Manal M Khan,** Professor, Department of Burns and Plastic Surgery, AIIMS Bhopal.

Rajnish Joshi
Dean (Academics)
AIIMS Bhopal

Place: AIIMS Bhopal
Date: 15/10/24

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Name of the Guide and Designation:

Dr. John A Santoshi,

Professor,

Department of Orthopaedics,

AIIMS, Bhopal.

डॉ. जॉन ए संतोषी/ Dr. John A. Santoshi
MS Orth, DNB Orth, F Hand Surg /Reg No.MP11493
प्राध्यापक/Professor
अस्थिरोग विभाग/Department of Orthopaedics
अखिल भारतीय आयुर्विज्ञान संस्थान भोपाल-462020
All India Institute of Medical Sciences Bhopal-462020

Name of the student

Dr. Shikha Malik,

Professor,

Department of Pediatrics,

AIIMS, Bhopal

डॉ. शिखा मलिक रजि. नं. M.P.-11453
प्राध्यापक एवं विभागाध्यक्ष
बाल्य एवं शिशु रोग विभाग
अखिल भारतीय आयुर्विज्ञान संस्थान
साकेत नगर, भोपाल-462020 (म.प्र.)

Names of co-guides and designation:

Dr. Prateek Behera,

Additional Professor,

Department of Orthopaedics,

डॉ. प्रतीक बेहेरा / Dr. PRATEEK BEHERA
Reg.No.16818/08 MS, DNB, MNAMS
अपर प्राध्यापक/Additional Professor
अस्थिरोग विभाग/Department of Orthopaedics
अखिल भारतीय आयुर्विज्ञान संस्थान भोपाल-462020
All India Institute of Medical Sciences Bhopal-462020

Dr. Manal M Khan

Professor,

Department of Burns and Plastic Surgery,

AIIMS Bhopal

डॉ. मनाल एम खान / Dr. MANAL M. KHAN
M.S.,M.Ch.(Plastic & Reconst. Surgery) Reg.No.25149
प्राध्यापक एवं विभागाध्यक्ष/Professor & HOD
बर्न्स एण्ड प्लास्टिक सर्जरी विभाग/Deptt. of Burns & Plastic Surgery
अखिल भारतीय आयुर्विज्ञान संस्थान भोपाल-462020
All India Institute of Medical Sciences Bhopal-462020

Place: AIIMS, Bhopal

Date: 15.10.2024

All India Institute of Medical Sciences, Bhopal

Institutional Human Ethics Committee-Student Research

MS Thesis Submission

By

Dr. Priya Maheshwari

Junior Resident (ACADEMICS)

(Reg No: 2200437M02P31010)

Department of Orthopaedics.

Title: " SPECTRUM OF PATIENTS WITH CONGENITAL LIMB DEFECTS PRESENTING AT A TERTIARY HEALTH CARE HOSPITAL "

Guide

Dr. John A Santoshi

Professor,

Department of Orthopaedics,

AIIMS, Bhopal.

डॉ. जॉन ए संतोषी / Dr. John A. Santoshi
MS Orth, DNB Orth, F Hand Surg. / Reg No.MP11493
प्राध्यापक / Professor
अस्थिरोग विभाग / Department of Orthopaedics
अखिल भारतीय आयुर्विज्ञान संस्थान भोपाल-462020
All India Institute of Medical Sciences Bhopal-462020

Co-Guides

Dr. Prateek Behera,

Additional Professor,

Department of Orthopaedics,

AIIMS, Bhopal

डॉ. प्रतीक बेहेरा / Dr. PRATEEK BEHERA
Reg.No.16818/08 MS, DNB, MNAMS
अपर प्राध्यापक / Additional Professor
अस्थिरोग विभाग / Department of Orthopaedics
अखिल भारतीय आयुर्विज्ञान संस्थान भोपाल-462020
All India Institute of Medical Sciences Bhopal-462020

Dr. Shikha Malik,

Professor,

Department of Pediatrics,

AIIMS, Bhopal

डॉ. शिखा मलिक / Dr. Shikha Malik
रजि. नं. MP-11493 24
प्राध्यापक एवं विभागाध्यक्ष
बाल्य एवं शिशु रोग विभाग
अखिल भारतीय आयुर्विज्ञान संस्थान
साकेत नगर, भोपाल-462020 (म.प्र.)

Dr. Manal M Khan

Professor,

Department of Burns and Plastic Surgery,

AIIMS, Bhopal

डॉ. मनाल एम. खान / Dr. MANAL M. KHAN
M.S. M.Ch. (Plastic & Reconst. Surgery) Reg.No.25149
प्राध्यापक एवं विभागाध्यक्ष / Professor & HOD
बर्न एण्ड प्लास्टिक सर्जरी विभाग / Deptt. of Burns & Plastic Surgery
अखिल भारतीय आयुर्विज्ञान संस्थान भोपाल-462020
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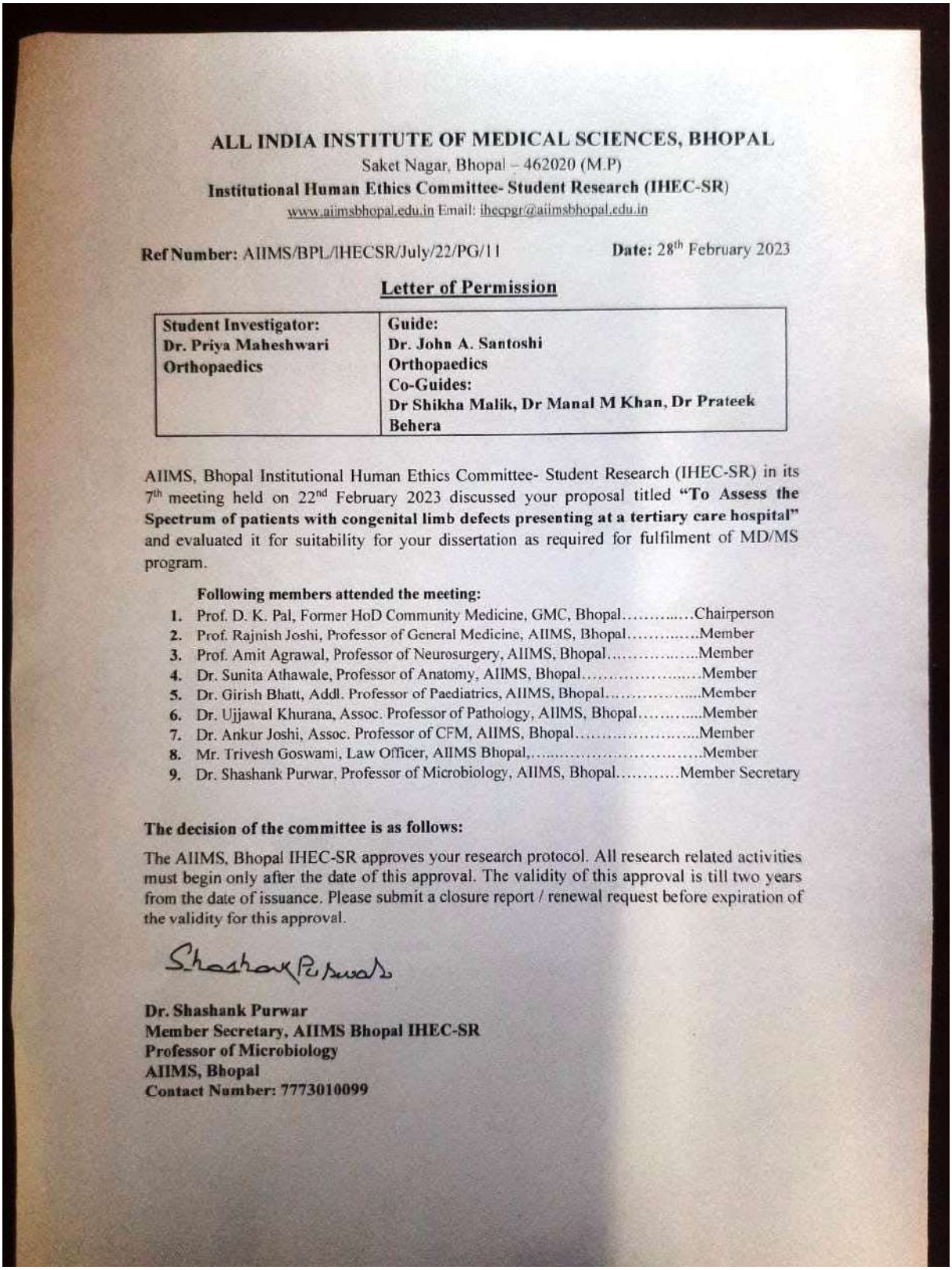

ALL INDIA INSTITUTE OF MEDICAL SCIENCES, BHOPAL

Saket Nagar, Bhopal – 462020 (M.P)

Institutional Human Ethics Committee- Student Research (IHEC-SR)

www.aiimsbhopal.edu.in Email: ihecpgr@aiimsbhopal.edu.in

Ref Number: AIIMS/BPL/IHECSR/July/22/PG/11 **Date:** 28th February 2023

Letter of Permission

Student Investigator: **Dr. Priya Maheshwari** **Orthopaedics**	**Guide:** **Dr. John A. Santoshi** **Orthopaedics** **Co-Guides:** **Dr Shikha Malik, Dr Manal M Khan, Dr Prateek Behera**

AIIMS, Bhopal Institutional Human Ethics Committee- Student Research (IHEC-SR) in its 7th meeting held on 22nd February 2023 discussed your proposal titled **"To Assess the Spectrum of patients with congenital limb defects presenting at a tertiary care hospital"** and evaluated it for suitability for your dissertation as required for fulfilment of MD/MS program.

Following members attended the meeting:

1. Prof. D. K. Pal, Former HoD Community Medicine, GMC, Bhopal……Chairperson
2. Prof. Rajnish Joshi, Professor of General Medicine, AIIMS, Bhopal……Member
3. Prof. Amit Agrawal, Professor of Neurosurgery, AIIMS, Bhopal……Member
4. Dr. Sunita Athawale, Professor of Anatomy, AIIMS, Bhopal……Member
5. Dr. Girish Bhatt, Addl. Professor of Paediatrics, AIIMS, Bhopal……Member
6. Dr. Ujjawal Khurana, Assoc. Professor of Pathology, AIIMS, Bhopal……Member
7. Dr. Ankur Joshi, Assoc. Professor of CFM, AIIMS, Bhopal……Member
8. Mr. Trivesh Goswami, Law Officer, AIIMS Bhopal,……Member
9. Dr. Shashank Purwar, Professor of Microbiology, AIIMS, Bhopal……Member Secretary

The decision of the committee is as follows:

The AIIMS, Bhopal IHEC-SR approves your research protocol. All research related activities must begin only after the date of this approval. The validity of this approval is till two years from the date of issuance. Please submit a closure report / renewal request before expiration of the validity for this approval.

Shashank Purwar

Dr. Shashank Purwar
Member Secretary, AIIMS Bhopal IHEC-SR
Professor of Microbiology
AIIMS, Bhopal
Contact Number: 7773010099

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Dr. Priya Maheshwari

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LIST OF ABBREVIATIONS

CLD	congenital limb defect
AER	Apical ectodermal ridge
PIP	Proximal interphalangeal
FDS	Flexor digitorum superficialis
CCBS	Congenital constriction band
Syndrome PFFD	Proximal focal femoral deficiency
RCH	Radial club hand
CTEV	Congenital talipes equinovarus
CVT	Congenital vertical talus
CPT	Congenital tibial pseudoarthrosis
FH	Fibular hemimelia
DDH	Developmental dysplasia of hip
ZPA	Zone of polarizing activity
SHH	Sonic hedgehog pathway
FGF	Fibroblast growth factor
BMP	Bone morphogenic protein
WHO	World health organization
GIT	Gastrointestinal tract

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INTRODUCTION

Interest in congenital malformations dates back to ancient times. William Harvey sought to understand the causes of these malformations and concluded that they arise from developmental disturbances. Congenital malformations are defined as structural abnormalities present at birth, resulting from improper development. These malformations arise from the complex interplay of genetic and environmental factors, with multifactorial inheritance thought to be the underlying cause of most common malformations. Various teratogens can impact the developing fetus during intrauterine life, leading to permanent postnatal damage, changes in morphology, or functional impairments. Teratogens include medications like thalidomide, infections such as rubella, radiation, chromosomal disorders, and nutritional deficiencies. Additionally, certain maternal factors—such as maternal age, parity, antenatal illnesses, and medication use—as well as paternal factors like age, education level, smoking, chronic illnesses, or being a carrier of congenital diseases, have been associated with these congenital defects. The overall incidence of congenital malformations is approximately 2% of all births. [1]

In India, interest in congenital malformations began to grow in the 1960s, following the publication of a study that analyzed data on various congenital malformations collected from multiple maternity hospitals in Mumbai.[2] Soon, additional studies emerged from various regions of the country, addressing different aspects of congenital malformations. Abnormalities of the musculoskeletal system are commonly reported, with an incidence of up to 9.69 cases per 1,000 births. Congenital limb defects (CLDs) are the second most prevalent type of birth defect.[1] According to the Global Report on Birth Defects, approximately 7.9 million births each year worldwide are affected by serious birth defects, with 94% occurring in middle- and low-income countries. A report from a joint meeting of the WHO and MOD states that birth defects contribute to 7% of

all neonatal mortality and account for 3.3 million deaths in children under five. Despite having the highest number of children with birth defects, congenital limb defects (CLDs) are not prioritized as a significant health issue in India. The prevalence of birth defects in India ranges from 61 to 69.9 per 1,000 live births. As the second most populous country in the world, India has a high birth rate but lacks a proper screening program for identifying CLDs. Medical attention for CLDs is critical from birth, yet this is often inadequate in developing countries like India, where advanced medical facilities are primarily concentrated in urban areas.

Congenital defects are a significant cause of mortality and morbidity. CLDs occur when the normal intrauterine development of a limb is disrupted. Children and adolescents with congenital limb defects often appear visibly different from their peers and may face limitations in their activities, depending on the severity of the defect. There is a scarcity of studies on congenital limb defects from India, particularly in central India, and limited data on patient-reported outcome measures for these individuals. Understanding the spectrum of patients with CLDs would facilitate comparative studies and illustrate changes and trends over time. This study aims to generate data that can assist treating physicians and policymakers in effectively planning and allocating resources for managing individuals with CLDs.

REVIEW OF LITERATURE

Embryology of Limb Development- By the end of the 4th week, limb buds appear as outgrowths from the ventrolateral body wall. These buds initially consist of a mesenchymal core derived from the lateral plate mesoderm, which gives rise to bones and connective tissue. Limb development follows a proximodistal pattern, guided by the apical ectodermal ridge (AER), a specialized region rich in stem cells that drives further limb growth.

By the 6th week, the terminal portions of the limb buds flatten to form handplates and footplates, separated from the proximal limb by a constriction. Another constriction appears in the proximal region, dividing the limb into two segments. Fingers and toes emerge as cell death occurs in the AER, and the digits continue to develop under the influence of five segments of ridge ectoderm. Mesenchymal condensation forms cartilaginous digital rays, and cell death between these rays' results in free digits.

Lower limb development mirrors that of the upper limb, but lags by about 2-3 days. By the 7th week, the limbs undergo a rotation: the upper limbs rotate 90 degrees laterally, placing the extensor surfaces on the lateral and posterior sides, while the lower limbs rotate 90 degrees medially, positioning the extensor muscles anteriorly and the great toe medially.

During this process, mesenchymal cells differentiate into chondrocytes, and by the end of the 6th week, the first hyaline cartilage models of bones form. Joint formation occurs when chondrogenesis is halted and a joint interzone is induced, a process driven by the WNT14 pathway. Endochondral ossification of bones begins by the end of the embryonic period, with primary ossification centers forming in all long bones by the 12th week. At birth, the diaphyses are ossified, while the epiphyses remain cartilaginous. Ossification of the epiphyses follows, but a temporary cartilaginous growth plate persists between the diaphysis and

epiphysis, facilitating longitudinal bone growth [2]. The epiphyseal plate is crucial for the longitudinal growth of bones. In long bones, there are two epiphyseal plates, one at each end. However, in smaller bones like the phalanges, the epiphysis is present only at one end. In irregular bones, there can be one or multiple primary ossification centers.

MOLECULAR REGULATION OF LIMB DEVELOPMENT

The positioning of limbs along the craniocaudal axis is controlled by **HOX genes**, which are expressed in an overlapping pattern from head to toe, with certain genes having cranial boundaries. Misexpression of these genes can disrupt normal limb development. Once the craniocaudal positioning of the limb is established, the remaining axes—**proximodistal**, **anteroposterior**, and **dorsoventral**—must also be determined to guide proper limb growth [3]. Lateral mesoderm cells begin secreting **FGF-10**, which triggers limb outgrowth. Subsequently, **bone morphogenic proteins (BMP)** released by the ventral ectoderm promote the formation of the **apical ectodermal ridge (AER)** by regulating the homeobox gene **MSX2**. This process is followed by the expression of another gene called **radical fringe**, located in the dorsal half of the limb ectoderm, which inhibits the AER from extending to the distal tip of the limbs. As a result, the expression of the **Ser-2 gene** is induced, leading to the release of **FGF-4** and **FGF-8**, which are essential for maintaining the **progress zone**—a cluster of rapidly dividing cells adjacent to the ridge. The distal growth of these cells is influenced by the activity within the progress zone [4]. Cells located proximal to the progress zone exhibit a slowdown in activity, leading to a gradual halt in their division and differentiation. The patterning of the anteroposterior (AP) axis of the limb is governed by the zone of polarizing activity (ZPA), a group of cells that produce retinoic acid, which activates the Sonic Hedgehog (SHH) pathway. This pathway regulates growth along the AP axis, resulting in the expected arrangement of digits, with the thumb positioned

radially and the little finger located posteriorly. Misexpression within the SHH pathway can cause mirror-image duplication of limb structures.

Furthermore, the dorsoventral axis is regulated by **bone morphogenic proteins (BMPs)** in the ventral region, which induce the expression of the **EN1** molecule. EN1 inhibits **WNT7a**, confining its expression to the dorsal ectoderm. **WNT7a** promotes the release of **LMX1**, a homeodomain-containing protein also restricted to the dorsal mesenchyme. Additionally, WNT7a plays a role in maintaining SHH gene function, thus supporting anteroposterior growth as well. This indicates that these genes are interconnected through feedback loops.

Ultimately, it is the **HOX genes** that regulate the shape and size of bones. HOX gene expression is influenced by the combined actions of **SHH**, **FGFs**, and **WNT7a**, facilitating the development of the proximal (stylopod), middle (zeugopod), and distal (autopod) regions of the limb. Among these, **HOXA** and **HOXD** are key determinants in limb development.

Components of Morphology: Common Terms for Congenital Anomalies

A paper by Hennekam et al., focused on dysmorphology, has developed a standardized framework for terms used to describe human morphology. Below are the standardized terms defined for congenital anomalies [47].

Phenotype: All observable traits and functions of an individual, excluding the DNA sequence. Derived from Greek, it includes attributes at the cellular, tissue, and individual level, like behavior or physiology. Epigenetic factors can blur the line between phenotype and genotype.

Genotype: The primary DNA sequence of an individual, including nuclear and mitochondrial DNA. It can refer to the whole genome or a specific gene variant. Some factors, like histone marks, exist at the boundary between genotype and phenotype.

Morphologic Anomaly: An anatomical phenotype (microscopic or macroscopic) that significantly deviates from the reference population.

Subtypes:

- **Major Morphologic Anomaly**: Has significant health or appearance consequences now, in the past, or in the future.
- **Minor Morphologic Anomaly**: Minimal or no health impact but may slightly affect appearance.

Comment: The term comes from the Greek "anomalos," meaning “not normal.” Morphologic anomalies are distinct from physiological abnormalities, like hypernatremia. A substantial anomaly is typically found in <2.5% of the population or falls outside the normal range (> or <2 SD from the mean). Anomalies can

occur alone or as part of syndromes and may include various conditions such as malformations or dysplasias. Definitions of major and minor anomalies may vary, with major anomalies affecting function or social acceptability, while minor anomalies lack medical or cosmetic impact. Social attitudes toward physical differences complicate these classifications.

Replaced Terms: Defect, birth defect, deviation, anatomical abnormality.

Morphologic Variant: A mild anatomical difference representing a small deviation from the reference population, found in 2.5-10% of people.

Comment: Discontinuous traits (like ear tags) are always anomalies, while continuous traits (like hypermobile joints) may be variants or anomalies. In clinical genetics, "variant" can also describe a phenotype similar to, but distinct from, a known entity.

Replaces: Birth defect, deformity.

Malformation: A congenital, non-progressive anomaly in a single organ or body part due to altered development.

Comment: Originating from the Latin *malus* (bad) and *formare* (to shape), malformations arise during the embryonic period and remain static. They can be caused by genetic mutations or teratogens and may occur alone or as part of syndromes. Unlike deformations or disruptions, malformations result from intrinsic developmental issues.

Replaces: Birth defect, deformity.

Deformation: An altered shape or position of a body part caused by abnormal mechanical forces on a normal structure.

Comment: From the Latin *de-formare* ("to shape"), deformations can lead to loss of symmetry or abnormal alignment. They may occur before or after birth and are usually reversible if treated early. Deformations can arise from external or internal forces and should not be confused with "deformity" or

malformations, which have different causes.

Replaces: Deformity.

Disruption: A non-progressive congenital anomaly caused by the breakdown of a body structure that had normal developmental potential.

Comment: Derived from Latin *dis* (apart) and *rumpere* (tear down), disruption refers to the destruction of normally developing body parts, typically occurring in utero. Causes include vascular issues, infections, teratogens, or mechanical forces. Disruptions may lead to abnormal growth and tissue damage and can occur in isolation or as part of syndromes. They should be distinguished from normal developmental processes like programmed cell death.

Dysplasia:Definition

Dysplasia refers to an abnormal change in tissue or cellular structure, occurring either before or after birth. It can affect an organ or tissue type due to ongoing changes in cellular makeup, organization, or function.

Key Points

- The term "dysplasia" comes from Greek, meaning "bad formation."
- Dysplasia may affect a part or the entirety of an organ.
- Unlike malformation or deformation, dysplasia often persists or worsens as the affected tissue grows.
- Causes include genetic factors or environmental exposures like teratogens.
- Dysplasia can occur in a single organ or across multiple systems and may be benign, progressive, or linked to cancer.

Synonym

Abnormal histiogenesis.

Syndrome: Definition

A syndrome is a set of anomalies, with at least one being structural, that are causally related.

Key Points

- Derived from Greek meaning "running together."
- In dysmorphology, syndromes include patterns of anomalies that are causally but not necessarily pathogenetically related.
- Examples include ciliopathies, rasopathies, and laminopathies.
- Chance co-occurrences of anomalies (false syndromes) are excluded.
- Syndromes can involve structural or functional abnormalities, including malformations, deformations, disruptions, sequences, or dysplasias.

Replaces

Spectrum.

Sequence: Definition

A sequence is one or more secondary morphological anomalies that arise from a single malformation, disruption, dysplasia, or deformation.

Key Points

- The term comes from the Latin word "sequentia," meaning "to follow."
- A sequence represents a chain of events resulting from an initial anomaly, unlike a syndrome, where multiple anomalies stem from a common cause, such as trisomy 21.
- Sequences can occur independently or as part of broader patterns, including syndromes.
- An example is the Pierre Robin sequence, where a small mandible leads to a protruding tongue, causing a cleft palate.

- Sequences may involve distinguishing primary, secondary, or tertiary effects, such as fetal akinesia leading to pulmonary hypoplasia.

Replaces

Anomalad, Complex, Cascade.

Association:Definition

An association is a pattern of anomalies, with at least two being morphological, that occur together more frequently than by chance, without an identified causal relationship.

Key Points

- The term originates from the Latin "associare," meaning "to make an ally."
- Associations may evolve into distinct syndromic entities as more information becomes available.
- Identifying associations can encourage clinicians to look for related anomalies that could be important for patient health and help develop differential diagnoses.
- Most associations are heterogeneous, often overlapping with multiple syndromes.
- Recognized examples include VACTERL/VATER (Vertebral malformations, Anal atresia, Cardiac defects, etc.) and MURCS (Müllerian anomalies, Renal aplasia, Cervical thoracic somite anomaly).
- Some associations, like the former CHARGE association, have been reclassified as syndromes after identifying a causative gene.

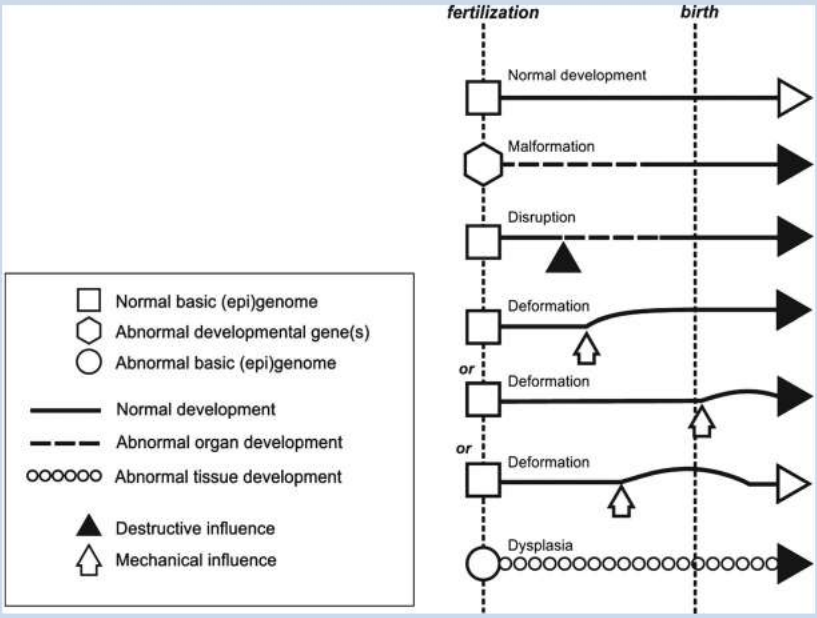


FIG. 1. Schematic representation of various types of morphologic anomalies compared to normal development.

CLINICAL CORRELATION OF EMBRYOLOGICAL DEVELOPMENT-

These limb defects are frequently associated with other anomalies, such as craniofacial, vertebral, and cardiac defects. Various types of limb deformities include **meromelia**, **phocomelia**, **amelia**, and **micromelia**. These anomalies are rare and are often linked to teratogenic drug exposure. For instance, **thalidomide** was widely prescribed to pregnant women in the 1960s to treat morning sickness, but it led to severe limb malformations, as well as cardiac and gastrointestinal anomalies. Although the drug was banned, it is now being reconsidered as a treatment for AIDS and cancer, raising concerns about a resurgence of limb defects associated with its use. The most critical period for teratogens to cause significant harm is during the 4th to 5th week of gestation. Another type of defect involves the digits. **Polydactyly**, characterized by the presence of extra rudimentary or fully developed digits, is typically autosomal dominant and often bilateral, though it can also be teratogen-

induced. **Ectrodactyly**, which results in the absence of a thumb, usually occurs unilaterally. **Syndactyly** occurs when the mesenchyme of the hand or foot plates fails to separate, causing digit fusion, which can sometimes extend to the phalanges.

Cleft hand or foot is characterized by an abnormal cleft between the 2nd and 4th metacarpals, often accompanied by the absence of the 3rd phalangeal and metacarpal bones. In some cases, the thumb may fuse with the index finger, and the ring finger may be fused with the little finger, giving the hand a lobster-like appearance. A mutation in the **HOXA13** gene is associated with **hand-foot-genital syndrome**, which is characterized by fused carpal bones and shortened digits [5]. Females may present with a bicornuate or didelphic uterus and an abnormal urethral orifice, while males often exhibit hypospadias. Mutations in the HOXD13 gene lead to a combination of syndactyly and polydactyly. Clubfoot, which can sometimes be hereditary, predominantly affects males and is characterized by the foot being turned inward, adducted, and plantar flexed. In cases of radial club hand, the radius may be absent or hypoplastic, and it can be associated with conditions like thrombocytopenia and craniosynostosis; the ulna is usually short and curved, with an absent or rudimentary thumb. Amniotic band syndrome, or congenital constriction bands, can lead to in-utero amputation of digits or limbs, though the exact cause remains unclear. Congenital hip dislocation, often seen in breech deliveries, results from abnormal positioning that causes laxity in the hip joint, leading to poorly formed hip joint development.

CONDITIONS TO BE INCLUDED IN THE STUDY

- **Polydactyly- Polydactyly**, also known as **hyperdactyly**, is the most common congenital anomaly affecting the hands and feet. It is characterized by the presence of an extra digit, which results from disruptions in the anteroposterior patterning during limb development. Polydactyly can occur in isolation (non-syndromic) or be associated with other birth defects, presenting as **syndromic polydactyly** [6]. Some patients may have a family history of polydactyly. **Syndromic polydactyly** can be associated with conditions such as **Diamond-Blackfan anemia**, **Fanconi anemia**, **Ellis-van Creveld syndrome**, **VACTERL syndrome**, and **trisomy 13** and **21**. Therefore, it is crucial to properly evaluate a child with polydactyly at birth. Epidemiological studies have shown that the **right upper limb** is most commonly affected. In a study by Finley et al., polydactyly was found to be **10 times more common in the Black population**. The occurrence of polydactyly is influenced by both **genetic** and **ethnic factors** [7]. The incidence of **pre-axial polydactyly** ranges from 0.08 to 1.4 per 1,000 live births, while **post-axial polydactyly** occurs in 1 to 2 per 1,000 live births. The prognosis for children with **isolated polydactyly** is generally excellent. However, when polydactyly is associated with syndromes, the prognosis may be less favorable.
- **Syndactyly-** Syndactyly is one of the most common congenital limb deformities, characterized by the fusion of certain fingers and/or toes. It can occur as an isolated condition or as part of more than 300 syndromic anomalies. **Syndactyly** shows significant clinical variability both between and within families. The phenotype can vary greatly even in the same individual, appearing as **unilateral** or **bilateral**, and **symmetrical** or **asymmetrical**. At least nine forms of non-syndromic syndactyly, along with several subtypes, have been identified. While most cases are inherited in an **autosomal dominant** manner, two **recessive** and **X-linked** forms have also been documented. The prevalence of syndactyly

is 3-10 per 10,000 live births. It can present as **partial**, **complete**, **cutaneous**, or **bony**, involving only the phalanges or extending to the metacarpal/metatarsal or even carpal/tarsal levels, and occasionally reaching the distal forearm or leg. The current classification of syndactyly is an adaptation and extension of the **Temtamy-McKusick** system, incorporating clinical, genetic, and molecular developments [52].

- **Macrodactyly- Macrodactyly** is a rare congenital condition characterized by the overgrowth of digits in the upper or lower limbs. Upton (2005) classified macrodactyly into four types: **Type I** involves macrodactyly with lipofibromatosis of a nerve, which can be either static or progressive; **Type II** is associated with **neurofibromatosis**; **Type III** presents with associated **hyperostosis**; and **Type IV** is linked to **hemihypertrophy**. Macrodactyly can also occur in conditions like **Beckwith-Wiedemann syndrome** and **Proteus syndrome**, or in isolation, where cosmetic concerns are a key factor in deciding onsurgicalintervention[8].

- **Clubfoot** - Commonly known as **clubfoot**, this condition is defined by the fixation of the foot in **adduction**, **supination**, and **varus**, along with associated soft tissue abnormalities. While the precise etiology is not fully understood, it is linked to various factors including **neurological**, **muscular**, **bony**, **connective tissue**, and **vascular mechanisms**. Milder cases are often attributed to abnormal **intrauterine posture**, and genetic factors play a role, with family history present in 24-50% of cases. In **clubfoot**, the foot is supinated, but the front of the foot is pronated relative to the back. The condition can be either **idiopathic** or **syndromic**. **Idiopathic clubfoot** is the most common type, typically occurring in isolation with normal upper limbs, and is 2 to 2.5 times more common in males than females. Syndromic cases, on the other hand, are associated with other anomalies, often arising with neurological or neuromuscular disorders such as **spina bifida** or **spinal muscular atrophies (SMAs)**. Idiopathic cases may also be linked to **joint laxity**, **congenital hip dysplasia**

(CDH), **tibial torsion**, and **ray anomalies of the foot** [9].

- **Congenital vertical talus- Congenital vertical talus (CVT)** is characterized by a **dorsal dislocation** of the **navicular bone**, resulting in **equinus** and **hindfoot valgus**. It is typically associated with multiple foot deformities, including **forefoot abduction** and **midfoot dorsiflexion**, primarily due to the fixed dorsal dislocation of the navicular bone over the talus. In 1913, Rocher coined the term "foot in piole" for this condition. CVT is also referred to as **reverse clubfoot**, **congenital valgus flatfoot**, **congenital convex pes valgus**, or **rocker bottom foot**. It may be associated with other conditions such as **cryptorchidism**, **microcephaly**, and **hypoxic-ischemic encephalopathy**. If left untreated, CVT can lead to severe, painful **callosities** and chronic foot and ankle pain. While traditional surgical management carries a high risk of complications, minimally invasive techniques have shown promising results in correcting deformities with limited soft tissue release. CVT is estimated to occur in **1 in 10,000 live births**. The valgus and equinus deformities in CVT are primarily caused by **contracture** of the **subtalar joint capsule**, **Achilles tendon**, and posterolateral ankle structures. **Dorsiflexion** and **abduction** at the forefoot and midfoot are due to contracture of muscles and tendons such as the **peroneus tertius**, **extensor digitorum longus**, **tibialis anterior**, and the **extensor hallucis longus and brevis**, as well as the **dorsal talonavicular capsule**. Additionally, **anterior subluxation** of the **peroneus longus** and **tibialis posterior tendons** contributes to ankle joint dorsiflexion [10].
- **Metatarsus adductus- Metatarsus adductus (MA)** occurs in **1 to 2 per 1000 live births** and is the most common congenital foot deformity in newborns. It presents as a curved or adducted **forefoot** with a normal **hindfoot**. Several factors contribute to its development, including 31**intrauterine compression**, **peroneal muscle weakness** with overactive **tibialis anterior** and **tibialis posterior**,

and **abnormal insertion of the tibialis posterior** on the 1st cuneiform. MA is associated with **developmental dysplasia of the hip (DDH)** in 1 to 5% of cases. Four types of MA are recognized: **simple**, **complex**, **skew foot**, and **complex skew foot**. Another classification by **Bleck** categorizes MA as **mild**, **moderate**, or **rigid**. Although the deformity may be present at birth, it often becomes more noticeable when the child begins to crawl, later leading to **in-toeing gait** and difficulty wearing shoes. In mild cases, the bisector line of the forefoot passes through the 3rd toe, while in moderate cases it passes between the 3rd and 4th toes, and in severe cases between the 4th and 5th toes. **Mild deformities** typically require no treatment, while **moderate to severe cases** may benefit from stretching, serial casting, or surgery if there is **pain**, difficulty walking, or cosmetic concerns [53].

- **Arthrogryposis –** The term “arthrogryposis” is derived from Greek, where "arthros" means joint and "gryposis" means crooked. Arthrogryposis can be classified into several categories:

  Amyoplasia: A muscle disorder that affects all four limbs.

  Distal arthrogryposis: An autosomal dominant condition that primarily affects the distal extremities.

  Arthrogryposis with central nervous system (CNS) etiology: Typically linked to abnormalities in the frontal lobe and often associated with conditions such as **microcephaly** or **hydrocephaly**.

- **Radial club hand -** Radial club hand refers to a range of complex congenital malformations affecting the radial side of the forearm. These malformations are relatively rare and can vary from a mildly hypoplastic thumb to a completely absent radius. Radial club hand is often associated with several syndromes, necessitating thorough evaluation. Mild cases can typically be managed nonoperatively, while severe cases usually require surgical intervention.

Managing radial club hand necessitates a collaborative approach involving an interprofessional team to evaluate and treat affected patients. The genetic basis of radial dysplasia is still not fully understood, but there is a longstanding suspicion that it may be linked to reduced fibroblast growth factor function due to mutations in the sonic hedgehog gene. Various potential causes for radial club hand have been proposed, including vascular insufficiency, intrauterine compression, environmental factors, drug exposure during pregnancy, and genetic influences.

Although radial dysplasia is relatively uncommon, it is the most frequently occurring type of congenital longitudinal deficiency. It is more prevalent in boys, with a ratio of 3:2. Estimates suggest that radial club hand occurs in about 1 in 55,000 births, although a study from Finland reported an incidence of approximately 1 in 5,000 live births. The condition is frequently associated with numerous congenital syndromes. In fact, only about one-third of patients with radial dysplasia present with an isolated upper extremity malformation, highlighting the importance of a comprehensive physical examination to detect associated congenital syndromes. Consultation with a clinical geneticist is advisable to identify any possible congenital syndrome associations. Some of the most commonly linked congenital syndromes include VACTERL association, VATER syndrome, Holt-Oram syndrome, TAR syndrome, and Fanconi anemia. To identify any associated conditions, spine radiographs, renal ultrasounds, complete blood counts, and echocardiograms are recommended [11]. Also if we discuss about limb defects, one of the diagnostic criteria for CHARGE syndrome, are being increasingly recognized. These anomalies include issues like polydactyly (extra fingers or toes), syndactyly (fused digits), and radial ray defects (such as missing or underdeveloped thumbs) [37, 39). The variability in these malformations reflects the syndrome's broad phenotypic spectrum.

The underlying cause is typically a mutation in the **CHD7 gene**, which plays a key role in regulating

chromatin structure and affects neural crest cells, essential for the development of the limbs and face [38, 42]. This common pathway explains why some patients with CHARGE syndrome have both craniofacial and limb anomalies. The frequency of limb malformations varies across studies. For example, **Sanlaville and Verloes (2007)** reported that about 15% of CHARGE syndrome patients exhibit limb anomalies [37], while **Hudson et al. (2012)** indicated that these may be more common but underreported [44]. Limb defects can have a significant impact on a patient's physical abilities and quality of life. For example, radial ray defects can lead to challenges with grasping or fine motor skills. Early orthopedic interventions, such as corrective surgeries, along with physical and occupational therapy, are often necessary to improve function and mobility [40,41]. The inclusion of limb defects in the phenotype highlights the need for flexibility in diagnosing and managing CHARGE syndrome, as the condition's clinical features continue to expand beyond the classical CHARGE acronym [43]. Bayne and Klug classified radial dysplasia into four categories based on the extent of radius involvement. Type I refers to mild radial shortening, while Type II is characterized by radius hypoplasia. Type III indicates cases with partial absence of the radius, and Type IV is defined by complete absence of the radius. James et al. later refined this classification by introducing Types N and 0 to account for isolated carpal anomalies in patients with a normally sized distal radius. Types III and IV are the most prevalent and are associated with the highest degree of wrist radial deviation. Long-term studies have indicated that although children with radial dysplasia may experience significant limitations in strength and joint mobility, they generally face few, if any, restrictions in their daily activities.

- **Ulnar club hand-** Ulnar Club Hand is a congenital deformity of the upper extremity marked

by a deficiency in the ulna and/or the ulnar-sided carpal structures. It can be associated with various orthopedic conditions, including:

- Scoliosis
- Phocomelia
- Proximal Femoral Focal Deficiency (PFFD)
- Fibular hemimelia
- Multiple hand abnormalities
- Nearly all patients exhibit an absence of ulnar-sided digits. This condition is classified according to Bayne's classification system. The etiology of Ulnar Club Hand is similar to that of Radial Club Hand [54].

Cleft hand – Cleft hand, also known as ectrodactyly or informally referred to as "split hand," is characterized by a central longitudinal deficiency that results in the underdevelopment of bone and soft tissues in the central fingers, specifically the index, middle, and ring fingers. This congenital disorder is present at birth and often has a positive family history. Cleft hand can be classified into two types: typical and atypical, with the atypical form frequently associated with Poland's syndrome. A distinctive "V-shaped" cleft is typically observed in the hand, exhibiting varying degrees of deformity. In most cases, the phalanges of the affected fingers are absent, while the metacarpals remain present. This condition is generally bilateral [12].

- **Camptodactyly -** Camptodactyly is a relatively uncommon hand anomaly characterized by varying degrees of congenital or acquired flexion contracture at the proximal

interphalangeal (PIP) joint of the fingers, which can occur unilaterally or bilaterally. The fifth finger is most commonly affected, and the condition can manifest as an isolated anomaly or be associated with syndromes. Various potential causes have been identified, including abnormal lumbricals, a shortened flexor digitorum superficialis (FDS) often accompanied by skin shortening, tight fascial bands, a deficient dorsal central slip extensor mechanism, and alterations in the distal interphalangeal or metacarpophalangeal joints. Camptodactyly can present early or later in life and is typically inherited in an autosomal dominant manner. While this condition often does not result in functional impairment, patients may seek medical attention primarily due to concerns about cosmetic appearance or requirements for military service. Treatment options vary according to the severity of the condition and may include nonoperative approaches, such as splinting or stretching exercises, as well as surgical interventions like tendon release, fascial band release, muscle transfer, and tenotomy.

● **Clinodactyly-** Clinodactyly is characterized by a coronal angulation of the affected digit exceeding 10 degrees. This curvature results from the abnormal trapezoidal or triangular shape of one or more phalanges, leading to malalignment at the interphalangeal joint. The irregular shape causes asymmetric longitudinal growth, deviating from the normal axis of the finger, which creates the visible curvature. Clinodactyly can manifest in syndromic, familial, or sporadic forms and typically follows an autosomal dominant inheritance pattern, most frequently affecting the middle phalanx of the small finger. Studies also note involvement of the proximal phalanx of the index finger and thumb. Although the exact incidence is unclear, reported rates range from 1% to 19.5%. It is often bilateral, with males more commonly affected. Clinodactyly is associated with several syndromes, including Rubinstein-Taybi syndrome, Cenani-Lenz

syndactyly, Klinefelter syndrome, Turner syndrome, Fanconi anemia, and occurs in up to 25% of children with Down syndrome. Radiographs may reveal a C-shaped physis or delta phalanx. [13]

Congenital Constriction Band Syndrome (CCBS), also referred to as **amniotic band syndrome**, is a group of fetal malformations caused by fibrous bands that constrict various parts of the fetus in utero, leading to deformities, malformations, or disruptions. The syndrome occurs sporadically with an estimated incidence of approximately 1 in 15,000 live births. It is often characterized by soft tissue constriction, typically involving the deep fascia around the limbs at birth. Despite many theories attempting to explain its cause, the precise etiology remains unknown.

Clubfoot deformity has been reported in 12-56% of patients with CCBS, with the affected foot either occurring below an ipsilateral band or in a limb without a proximal band. Some studies have noted that clubfeet associated with CCBS are more rigid and resistant to nonoperative treatments. In earlier cases, extensive surgical release was commonly used to manage these cases. The combination of clubfoot and circumferential amniotic band syndrome is distinct from other congenital deformities due to the high risk of multiple relapses [55].

- **Phocomelia-** Phocomelia is a rare congenital disorder characterized by the absence of intermediate segments of the limbs, leading to hands or feet being directly attached to the trunk. This condition is notably linked to the teratogenic effects of the drug thalidomide, which was originally marketed for treating anxiety and morning sickness. Approximately 40% of infants

affected by thalidomide-related phocomelia died around the time of birth. The widespread occurrence of these birth defects significantly influenced social and political movements surrounding drug safety and regulation. Despite strict global regulation, several cases of thalidomide-induced phocomelia have been reported in the last three decades. In limb development, the apical ectodermal ridge (AER) forms at the distal tip of the limb bud. Its interaction with the underlying progress zone mesoderm is crucial for normal longitudinal growth and differentiation of proximal and distal limb structures. Any disruption to this relationship—whether due to vascular insufficiency, drug toxicity, or apoptosis—can result in phocomelia. Children born with this condition should also be evaluated for other associated abnormalities, particularly in the heart and gastrointestinal system. Syndromes associated with phocomelia include Roberts syndrome, thrombocytopenia-absent radius (TAR) syndrome, severe limb defect syndromes, vertebral hypersegmentation, and mirror polydactyly [14].

- **Amelia-** Amelia is a congenital condition characterized by the complete absence of skeletal components of a limb, resulting from the disruption or prevention of limb formation early in embryonic development, between 24 and 36 days after fertilization. Although generally considered a sporadic anomaly, over 50% of amelia cases are associated with significant malformations in other organ systems. Common accompanying malformations include cleft lip and/or palate, body wall defects, head abnormalities, and defects involving the neural tube, kidneys, and diaphragm.

Facial clefts may be associated with other facial anomalies, such as a small jaw or absent ears or nose. Body wall defects can cause internal organs to protrude through the abdominal wall, while head malformations can range from minor to severe, including near absence of the brain. The

diaphragm may be herniated or missing, and one or both kidneys may be underdeveloped or absent. Additional anomalies may affect the lungs, vertebrae, heart, genital system, and anus. Severe growth deficiencies are often present before and after birth, and mental retardation may occur in survivors.

Several factors have been implicated in causing amelia, including teratogens such as thalidomide and alcohol, vascular disruptions (e.g., from amniotic bands), and maternal conditions like diabetes [15].

Proximal focal femoral deficiency - Proximal focal femoral deficiency (PFFD) is a rare congenital skeletal disorder characterized by the partial absence of the proximal femur, resulting in shortening of the entire lower limb. Clinically, it manifests as a shortened thigh that is flexed, externally rotated, and abducted. Although the precise cause of PFFD remains unclear, factors such as maternal diabetes, drug exposure, viral infections, and radiation have been suggested as potential contributors. The condition is usually sporadic, although a few familial cases have been reported.

PFFD typically affects one limb in 85% to 90% of cases, and it is often associated with other skeletal anomalies such as fibular hemimelia, pes equinovarus, oligodactyly, and tibial bone bending in up to 60% of affected individuals. Diagnosis is typically made in early childhood through conventional radiography, but the radiographic features of the condition evolve as the child grows [16].

- **Fibular hemimelia** - Fibular hemimelia (FH) is a congenital condition characterized by underdevelopment or absence of the fibular bone, often accompanied by hypoplasia or dysplasia of the tibia and varying degrees of foot abnormalities. These foot anomalies may include hypoplasia, dysplasia, or aplasia of certain parts of the foot. The presentation of FH spans a broad spectrum, from

mild cases with minimal limb length discrepancy to severe deformities involving the ankle and foot, with or without subtalar coalition, midfoot coalitions, and absent rays. Commonly associated conditions include knee ligament deficiencies, knee valgus deformity, and femoral hypoplasia, dysplasia, or partial aplasia. FH belongs to the same spectrum of deficiencies as congenital femoral deficiency and is classified as a postaxial deficiency, distinguishing it from preaxial deficiencies like tibial hemimelia. The exact cause of FH remains unknown, and it is typically not an inherited condition, with most affected children having no family history of similar birth defects. Children with FH typically face five major challenges: limb length discrepancy, foot and ankle deformities, and tibial deformity [56].

- **Tibial hemimelia**- Tibial hemimelia typically manifests as a shortened leg accompanied by deformities in the knee and ankle. The tibia may be underdeveloped, entirely absent, or consist of a non-ossified remnant that is not visible on X-rays. It is common for affected individuals to experience knee flexion contractures, instability due to absent collateral ligaments, and the potential absence of the patella and quadriceps extensor mechanism. Skin dimples may be present over the knee joint.

Additionally, the fibula can be dysplastic and is frequently found to be subluxated or dislocated either proximally or distally. The ankle often presents in a varus and equinus position, with the foot typically adducted and supinated. Hypoplasia of the big toe or the absence of certain rays may occur on the medial side of the foot. Duplications of toes, metatarsals, tarsals, fibulas, and femurs are also characteristic features. Diagnosis of tibial hemimelia can be made via prenatal ultrasound by 16 weeks of gestation, and about 30% of cases are bilateral. There have been reports suggesting possible parent-to-child transmission, as well as families with multiple affected siblings, indicating potential autosomal dominant and autosomal recessive inheritance patterns, with consanguinity also being a factor. It is hypothesized that the underlying pathology originates from mesoblastic sources rather than mechanical or traumatic causes. Interestingly, variable phenotypic expressions have been noted, including cases of

identical twins where only one was affected. Tibial hemimelia is associated with several syndromes, including Werner's syndrome, Langer–Giedion syndrome, Gollop–Wolfgang complex, and CHARGE syndrome. Approximately 60% of affected individuals have associated anomalies. Ipsilateral thigh and knee deformities can include congenital femoral deficiency, the absence of the patella or quadriceps mechanism, knee hyperextension or flexion, and a bifid femur. Common foot deformities include clubfoot, syndactyly, absent or duplicated toes, diplopodia, ectrodactyly, and various other limb malformations. Additionally, conditions such as cleft palate, hearing loss, cryptorchidism, pseudo-hermaphroditism, and hypospadias have been linked to tibial hemimelia. Unlike fibular hemimelia, where the ankle and foot are often in a valgus position, in tibial hemimelia, they are typically found in a varus position. It is essential to differentiate tibial hemimelia from congenital knee deficiencies associated with thrombocytopenia-absent-radius syndrome [18].

- **Sirenomelia-** Sirenomelia is an extremely rare and often fatal congenital condition marked by varying degrees of fusion of the lower limbs, as well as anomalies in the thoracolumbar spine, sacrococcygeal agenesis, and abnormalities in the genitourinary and anorectal systems. It represents one of the most severe forms of caudal regression syndrome. Key features of sirenomelia include the fusion of the lower extremities, the presence of a single umbilical artery, and a persistent vitelline artery. The exact molecular cause of sirenomelia is not well understood, but two primary hypotheses have been proposed to explain its development: the vascular steal hypothesis and the defective blastogenesis hypothesis. The vascular steal hypothesis suggests that limb fusion occurs due to inadequate blood flow and nutrient delivery to the caudal mesoderm, leading to the agenesis of midline structures and abnormal approximation of the lower limb fields. In contrast, the defective blastogenesis

hypothesis attributes the primary defect in caudal mesoderm development to teratogenic events during the gastrulation phase, which disrupts notochord formation and subsequent development. Environmental factors, such as maternal exposure to certain teratogens, may also play a role in the development of this condition. Unfortunately, sirenomelia is typically fatal within one to two days after birth due to complications related to the abnormal development and function of the kidneys and urinary bladder [19].

- **Congenital tibial bowing -** Bowing of the tibia refers to the bending of the tibial diaphysis, with the apex of the curve directed either anterolaterally, anteromedially, or posteromedially. Posteromedial bowing is a congenital condition that is frequently associated with a calcaneovalgus deformity of the same-side foot. The primary causes of this condition can be categorized as congenital factors, fibular hypoplasia, pseudoarthrosis of the tibia, Blount's disease, and other miscellaneous causes, which may include physiological factors, rickets, osteogenesis imperfecta, fibrous dysplasia, and post-traumatic effects.

- **Congenital tibial pseudoarthrosis (CPT)-** The condition is caused by pathological changes in the periosteum of the bone. McElvenny noted that a thickened, adherent periosteum constricts the tibia and fibula, resulting in atrophy, fractures, and pseudarthrosis. Boyd, along with Boyd and Sage, suggested that congenital pseudarthrosis of the tibia (CPT) arises from osteolytic fibromatosis based on their examination of amputated specimens. In a study involving ten patients with CPT, Bluth et al. proposed that the thickened periosteum is a result of myofibroblast overgrowth. More recent research has identified neural-like cells surrounding small periosteal vessels, leading to narrowing and obliteration of these vessels, which may cause hypoxia in the subperiosteal bone. This hypoxia can subsequently result in bone resorption, fractures, and non-union. **Cho et al.** demonstrated that periosteal cells in the

hamartoma of CPT exhibit decreased osteoblastic responses to BMP-2, while the osteoclastic activity of the periosteum was significantly elevated compared to control groups. Consequently, Tey concluded that the failure of healing and resorption of bone grafts in CPT is associated with increased osteoclastic activity and reduced osteoblastic activity in the CPT periosteum when compared to normal periosteum. Additionally, CPT is often associated with neurofibromatosis type 1 [20].

Aldo Rosano et al. conducted a study that pooled data from 11 birth defect registries under the International Clearinghouse for Birth Defects Monitoring Systems. They identified 666 infants born between 1983 and 1993 who had a non-syndromic limb defect along with at least one other major malformation, resulting in a rate of 12.9 per 100,000 population. To analyze the association patterns, the researchers utilized observed/expected ratios and log-linear models.

Their findings indicated that certain limb defects were associated with specific sets of malformations. For instance, preaxial limb defects were more commonly linked to microtia, esophageal atresia, anorectal atresia, heart defects, unilateral kidney dysgenesis, and some axial skeleton anomalies. Postaxial defects were associated with hypospadias, while transverse defects correlated with craniofacial anomalies, micrognathia, ring constrictions, and muscular defects. Intercalary defects were linked to omphalocele, split hand/foot to encephalocele, and amelia to anorectal atresia, omphalocele, severe genital defects, unilateral kidney dysgenesis, gastroschisis, and ring constriction.

Log-linear modeling revealed higher-order associations among some of these malformations.
The researchers employed the chi-square test to assess the heterogeneity of prevalence rates and used Poisson regression to analyze time trends. They also referenced previous work on observed versus expected (OE) ratios and log-linear models to further identify associations

[21]. They used the former method to study associations of up to three defects, and the latter for those of three or more. In the OE method, the expected frequency of a defect combination is a function of the frequency of the individual defects among all multi-malformed infants, and the strength of the association is measured by the od

ratio (OR) [Kallen and Winberg, 1969]. In loglinear modelling, we disregarded one-way effects (associations of two defects) because such effects critically depend on the choice of defects [Beaty et al., 1991]. In addition, because log-linear models work best with large sample sizes, we restricted their use to the study of the more frequent limb defects (preaxial and transverse) and the more frequent associated anomalies (oesophageal and anorectal atresia, cardiac defects, axial skeletal defects, and urinary tract defects; micrognathia, anorectal atresia, other axial skeletal defects, severe genital defects, severe craniofacial defects, and cleft palate). They used the partial x2 test to rank the strength of the log-linear models.

In a study conducted by **Divine Thampi et al.** in 2017, 12 children aged 6 to 15 years and their mothers participated between January and June of that year. The researchers conducted in-depth interviews to explore the experiences and perspectives of these children with congenital hand differences and their mothers. The interviews were transcribed verbatim and analyzed using manual coding followed by thematic analysis.

The children in the study indicated that they managed most of their daily activities with minimal involvement from their parents, demonstrating a degree of independence in their self-care by adapting to their disabilities. They reported receiving care and support from family members. Although they faced societal stigma, they coped by embracing their disabilities, aided by positive reinforcement from their parents. Most mothers and children noted that surgical interventions had

enhanced the physical functionality of their hands. Notably, all participants mentioned they did not rely on orthoses for daily tasks; any orthoses provided were primarily for maintaining correction of their deformities and were mostly used as night splints

E. Koskimies et al. conducted a hospital-based study involving 362 children with upper limb defects, utilizing data from the Finnish Register of Congenital Malformations to identify live births. They collected data regarding hospital care, specifically focusing on operative interventions, from the national hospital discharge register up until December 31, 2009. The results indicated that out of a total of 513 orthopedic procedures performed, 326 were related to upper limb defects, while 107 targeted lower limb issues, with the remainder pertaining to spinal conditions

Afieharo Igbibia Michael et al. conducted a cross-sectional retrospective study of 925 pediatric outpatients at the hand clinic of University College Hospital, a tertiary center in southwestern Nigeria. They utilized descriptive and inferential statistics, employing frequencies, student's t-test, and chi-square test for their analysis. The study revealed a prevalence rate of congenital hand anomalies of 13.2%. A total of 152 cases were identified, with the highest number occurring in 2012, accounting for 23 cases. The study showed a male predominance, with 66 male patients. Approximately 26.2% of the patients presented as neonates, while 29.5% were infants. The most common anomaly was failure of differentiation, followed by duplication, with syndactyly being the most frequent condition within the failure of differentiation category. No significant association was observed between the type of anomaly and gender or laterality, although there was a notable prevalence of bilaterality in cases of polydactyly and syndactyly

Aryasinghe et al. conducted a descriptive study based on hospital records of 1,222 live births over a six-

month period from December 2007 to June 2008, identifying 84 cases of congenital anomalies. This results in an anomalous birth rate of 68.7 per 1,000 live births, with musculoskeletal anomalies being the second most common at 28.6%, following genitourinary anomalies. The most frequently observed anomaly was talipes.

The study found a significant association between birth weight (p=0.005) and gender (p=0.001) and the occurrence of birth defects. In contrast, maternal age, gravidity, parity, history of previous abortions, VDRL and Hepatitis B status, gestational diabetes, pregnancy-induced hypertension, gestational age, maternal and infant blood groups, nationality, and consanguinity showed no statistically significant association with congenital anomalies. This research was conducted at Gulf Medical College Hospital and Research Centre, the primary teaching hospital in Ajman, United Arab Emirates

Ekaterina Vasluian et al. conducted a population-based study examining the birth prevalence of congenital limb defects (CLD) over a 30-year period from 1981 to 2010. The study found a prevalence rate of 21.1 per 1,000 live births. Among the 1,048 children with CLD, 55% were male, 57% had isolated defects, while 13% presented with multiple congenital anomalies, and 30% had recognized anomalies. Cases were identified following the guidelines of the EUROCAT central registry, which employs the ICD-9 classification. The anatomical categories included polydactyly, syndactyly, reduction defects (transverse, longitudinal, intercalary, and central), as well as split hand and split foot

Importance of the proposed study in the current context

CLD is the second most common type of anomaly in children and is one of the leading causes of disability worldwide if or not associated with syndromes. Most common cases of CLD include those with polydactyly, syndactyly, amelia etc. It has been observed that, with better control of nutritional deficiencies and infections, CLDs have emerged as a sad cause for increasing peri-natal mortality. Very few studies have been conducted in India which include children with CLDs and hence the data available regarding the prevalence and distribution of the same is not enough. This study will help in estimating the prevalence of children with such defects.

AIMS AND OBJECTIVES

AIM OF THE STUDY:

- To assess the spectrum of patients with congenital limb defects presenting at a tertiary care hospital.

Objective:

Primary Objective

1. To find the distribution of various congenital limb defects presenting at a tertiary care hospital.

Secondary Objectives

1. To assess the health seeking behavior of the parent/patient regarding the congenital limb defect

1.1 Quality of life assessment of patients with congenital limb defects using patient-/parent- reported scoring system.

1.2 To do the cost analysis of treatment in case the patient undergoes/ has undergone corrective surgery.

MATERIALS AND METHODOLOGY

- **Study design:** hospital based observational study

- **Sample size-** we aimed to enroll around 120 patients. This number was based on the average number of patients of congenital limb defects seen in the departments of Orthopaedics, Paediatrics, Burns & Plastic surgery at our institution. During the time of calculation, around 80 patients with congenital limb defects were seen every year in the three departments involved in the study.

- **Place of the study**- Data was collected as per the proforma with consent from the patients/parents attending Orthopaedics, Paediatrics, and Burns & Plastic surgery departments of the institution.

- **Time period** – March 2023- September 2024

- **Study Population:** Patients seen in the departments of Orthopaedics, Paediatrics, and Burns & Plastic Surgery who had congenital limb defects were included. Patients who sought hospital care for unrelated issues and had congenital limb defects were subsequently enrolled in the study as part of the congenital limb defect cohort

Inclusion criteria

1. Patients with congenital limb defects
2. Patients/ parents who gave consent to participate in the study

Methodology:

The participants were recruited from the departments of Orthopaedics, Paediatrics, and Burns & Plastic surgery of our institution. The patients who were diagnosed cases of congenital limb defects were enrolled. The written informed consent from patients/ parents was obtained.

Detailed evaluation of all the patients including history, general examination, local examination, and investigations in the form of hematological parameters, radiographs and special tests/ imaging if any was taken into account.

- **PATIENT HISTORY:**
  - Name
  - Age
  - Age of the parents at the time of conception
  - Sex
  - Address / Contact details
  - Chief Complaints
  - Birth history, if known

- Famly history
- Treatment history
- Pedigree chart
- Consanguinity
- Gestational age at which the child is born, if known
- Risk factors – drugs/ smoking/ alcohol/ TORCH any other significant risk history if details are available
- Additional remarks from the patient and attendant
- GENERAL EXAMINATION:
- LOCAL EXAMINATION:
- Type of anomaly-
- Inspection
- Palpation
- Measurements

- **INVESTIGATIONS:** (done as part of routine work up of patient)

1. Radiographs of the affected part in two orthogonal views preferably AP, Lateral.
2. Genetic screening, if done
3. Haematological markers
4. 2D ECHO, USG abdomen, USG KUB, Cystourethrogram, barium studies.

Design and Ethical statement:

- We conduct an observational study in the departments of Orthopaedics, Paediatrics, Burns & Plastic surgery, AIIMS Bhopal, where patients with congenital limb defects will be observed and enrolled.
- All study related procedures will commence after due clearance from the Ethics committee (IHEC-PGR).
- All data would be collected from departments, Department of Orthopaedics, Paediatrics and Burns and Plastic Surgery AIIMS, Bhopal.
- Informed written consent will be taken from patients participating in the study.

OBSERVATION AND RESULTS

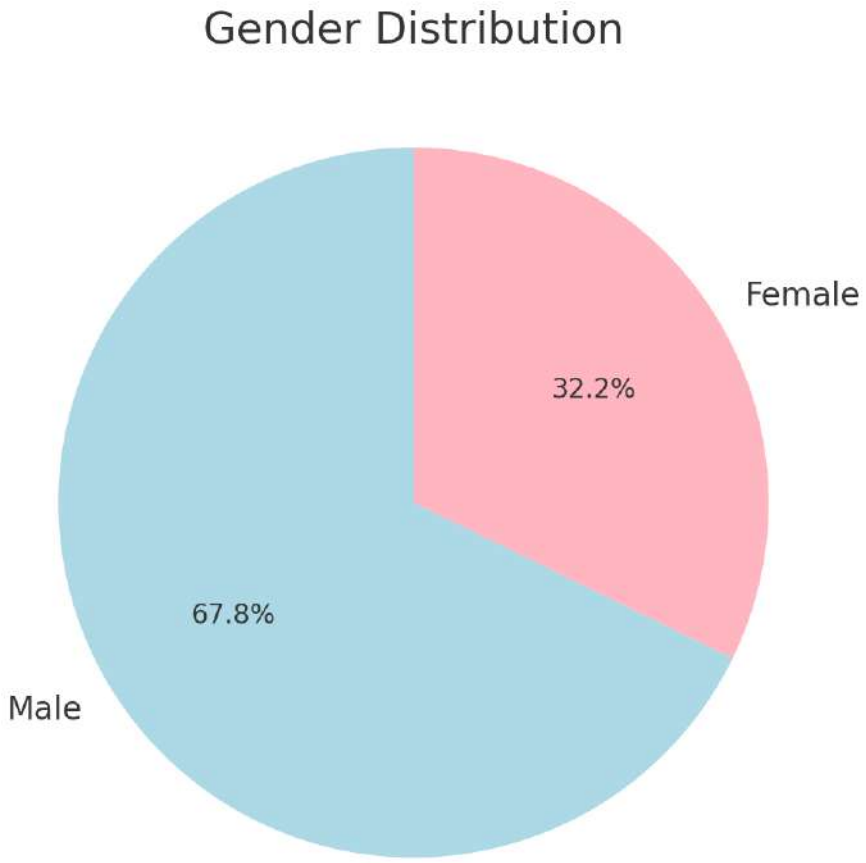
In this observational study, we assessed 121 patients with congenital limb defects. The study was conducted at AIIMS Bhopal from March 2023-September 2024.

Patients with CLDs who presented to the Department of Orthopedics, BPS and Pediatrics, AIIMS Bhopal after inclusion criteria were enrolled during the study period. March 2023-September 2024

Demographic and Diagnostic Analysis of Deformities in Study Population

Gender Distribution:

The study consisted of 121 patients, out of which 81 (67.5%) were male, and 40 (32.5%) were female, as illustrated in the gender distribution chart. This significant male preponderance highlights a greater occurrence of deformities in males as compared to females, potentially indicating a higher genetic or environmental predisposition among males.



pie chart showing gender distribution

Diagnosis Distribution:

The spectrum of congenital deformities in this study is varied, with several types of deformities identified among the patients. The most prevalent condition observed was clubfoot, affecting 56 patients. This condition is further presented in 21 patients bilaterally and 35 patients unilaterally. Additionally, 4 patients presented with Arthrogryposis Multiplex Congenita, and 2 patients had Congenital Constriction Band Syndrome as associated conditions.

Other significant deformities included:

- Tibia Hemimelia was observed in 3 patients, with 1 case being syndromic, associated with

bilateral radial club hand and mirror foot deformity.

- Radial club hand was present in 7 patients. associations were seen including syndromes like CHARGE syndrome (2), Holt Oram syndrome (1), Affection with TORCH infection in mother (1), bilateral radial club hand with hypoplastic lungs with brochiectasis(1)

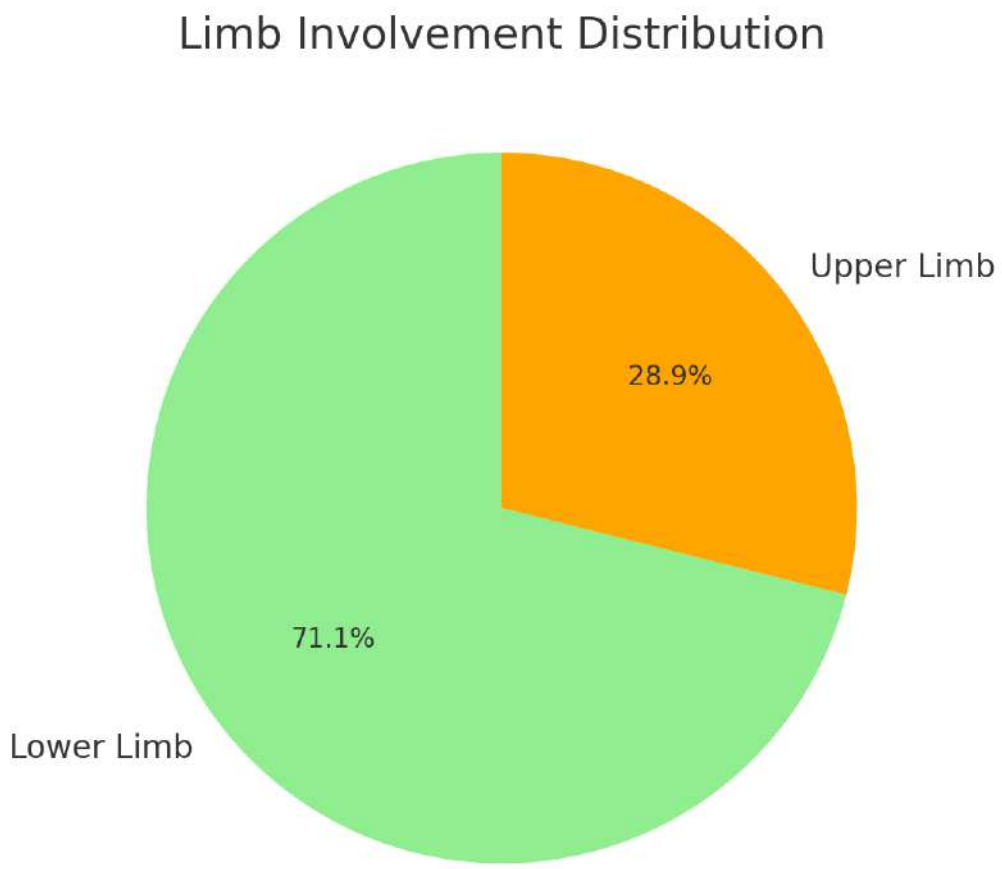
Additional deformities identified included:

- Syndactyly affecting 8 patients, with the most common fusion being between the middle and ring fingers.
- Polydactyly was diagnosed in 16 patients, with pre-axial involvement in 6 cases and post-axial in 4 cases for the hands, and an even split of pre-axial and post-axial types in the feet (3 cases each).
- Synpolydactyly was observed in 2 patients.
- Camptodactyly was observed in 2 patients

The study also identified rarer conditions such as proximal focal femoral deficiency (2 patients), brachydactyly (4 patients), and congenital vertical talus (3 patients), among others.

Limb Involvement Distribution:

Of the 121 patients analyzed, 86 patients (71.7%) had deformities affecting the lower limbs, while 35 patients (28.3%) had upper limb deformities. This substantial difference indicates a higher prevalence of lower limb involvement in the congenital deformities treated in this study. The pie chart on limb involvement distribution clearly emphasizes this imbalance.



pie chart showing distribution of CLDs by limb

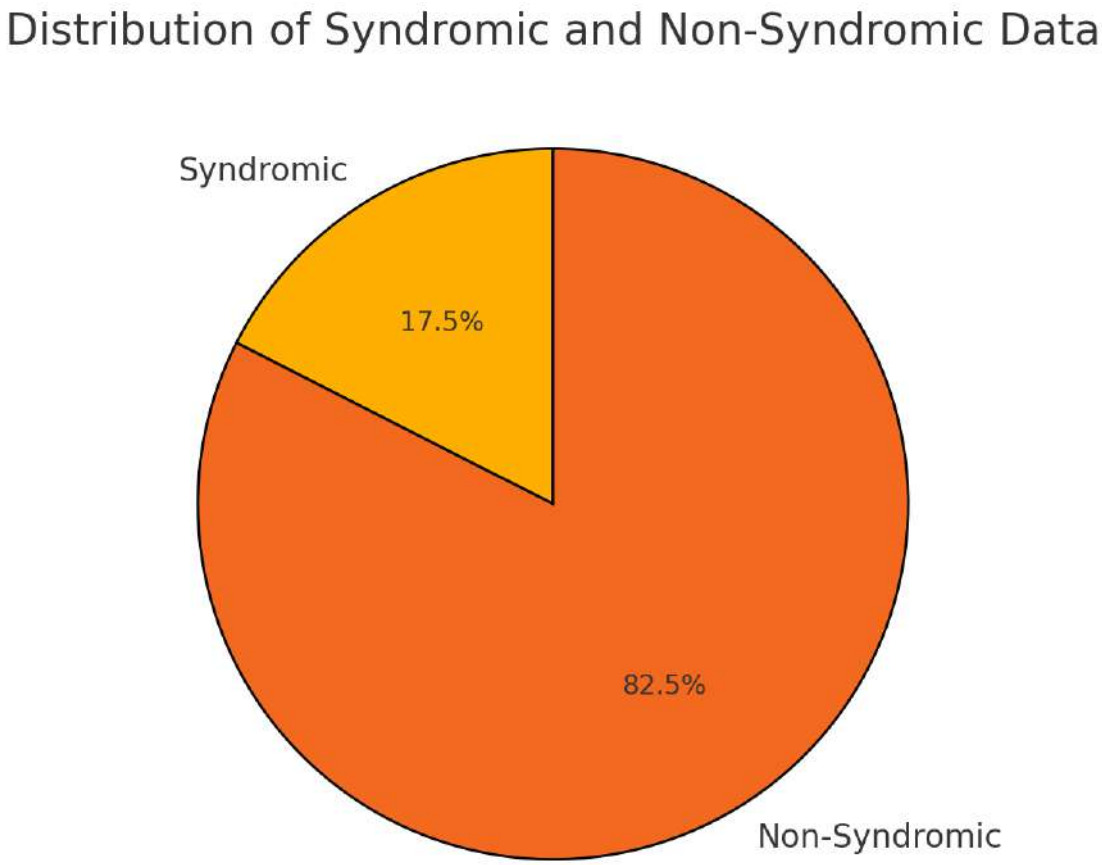
Age Group Distribution:

The age group distribution was another important factor in the study. Of the 121 patients, 19 (15%) were aged 18 years or older, while the majority, 102 patients (85%), were younger than 18. This finding suggests that most deformities were identified and treated in pediatric patients, with a smaller proportion of adults undergoing treatment for congenital conditions, possibly reflecting a trend of earlier diagnosis and intervention in younger individuals.

Key Associations with Syndromic Conditions:

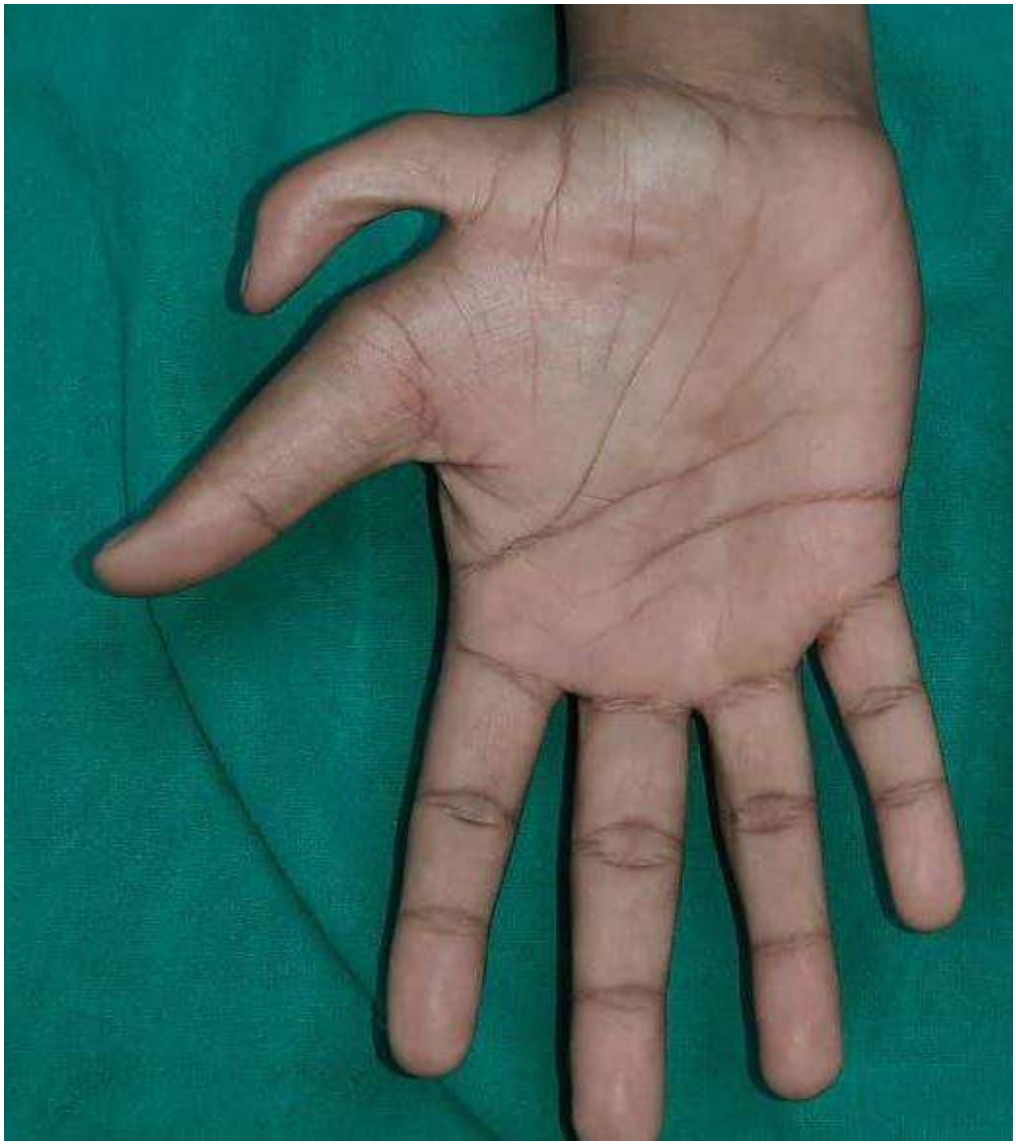
Several deformities were noted to be associated with specific syndromes:

- AMC and CCBS were notably found in conjunction with clubfoot.
- AMC and CCBS were also seen as separate entities.
- A single patient with bilateral radial club hand also presented with bilateral hypoplastic lung disease and bronchiectasis, emphasizing the syndromic nature of these deformities.
- Children with Holt Oram syndrome, CHARGE syndrome, Freeman Sheldon syndrome, congenital pterygium syndrome, Larsen syndrome and congenital rubella syndrome with immunodeficiency were also seen.
- Other complex associations included a case of syndromic bilateral radial club hand co-occurring with tibia hemimelia and mirror foot deformity.



Pie chart showing syndromic v/s non-syndromic distribution

Here are some of the clinical images of the patients captured during the study period.



Case of pre-axial polydactyly-right side

Child with right sided radial club hand

Syndromic child with tibia hemimelia right side with mirror foot

Child with right clubfoot

Child with right clubfoot

CPT of right side

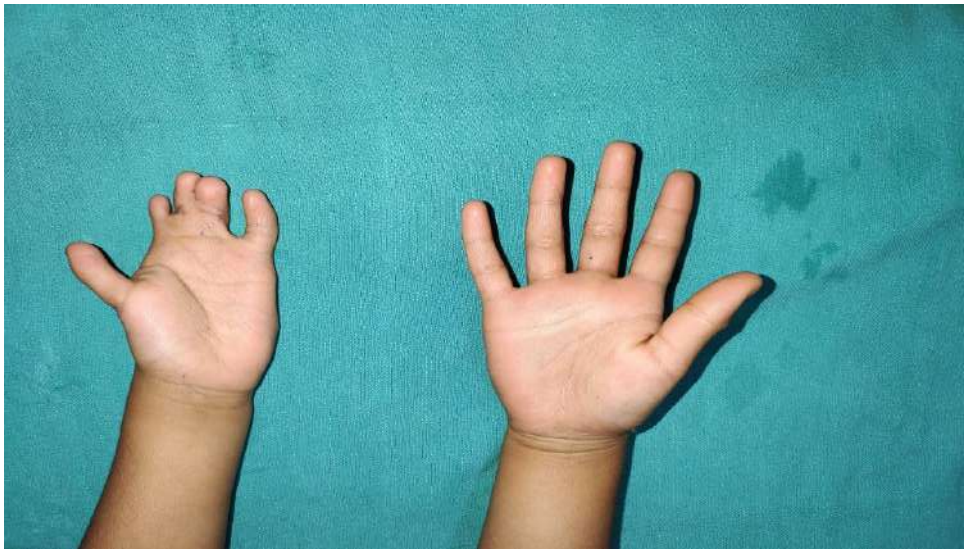
Child with neurofibromatosis type 1 with macrodactyly of right ring finger

Radio-ulnar synostosis- right side

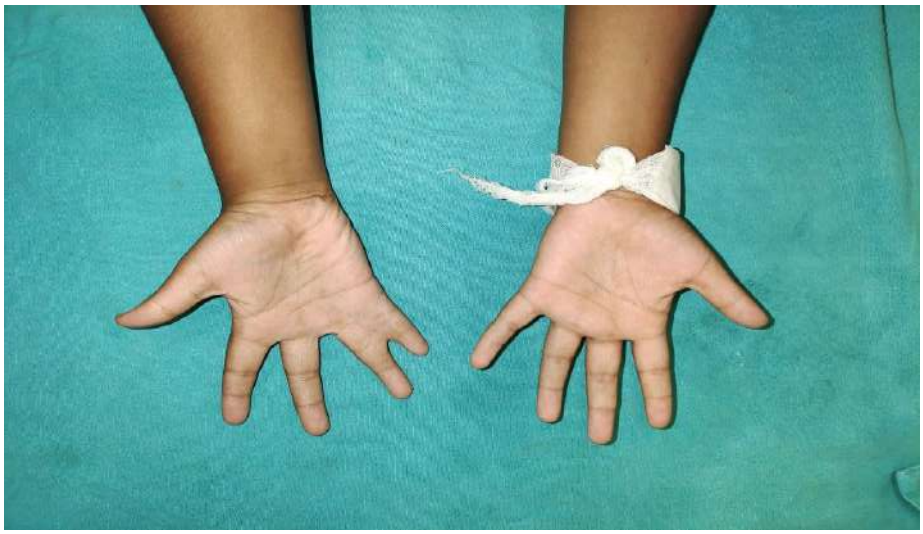
CCBS

Cleft hand right side

Synpolydactyly of feet



Synbrachydactyly



Syndactyly

Thumb hypoplasia

DISCUSSION

Congenital limb defects encompass a wide range of malformations that can significantly impact the physical and psychosocial well-being of affected individuals. Our study examined the demographic and diagnostic spectrum of congenital limb defects in a study population of 120 patients.

1. Demographic Analysis

Gender Distribution

We found out that 67.5% of the patients were male (81 patients), compared to 32.5% female (39 patients). This notable male predominance is consistent with findings in other studies, suggesting that congenital limb defects may have a genetic or environmental predisposition that favors males [22] [23]. For instance, an Indian study by **Gupta et al. (2015)** found similar trends, where 65% of their patient cohort with congenital limb defects were male, indicating a persistent pattern across different populations [24].

Age Group Distribution

The age distribution revealed that 85% (102 patients) were under 18 years old, while only 15% (18 patients) were aged 18 and older. This finding aligns with trends noted in international literature, suggesting that congenital deformities are primarily diagnosed and treated in the pediatric population [25]. A study by **Aroojis et al. (2020)** reported that early detection and intervention in congenital limb deformities lead to significantly better functional outcomes, underscoring the importance of pediatric orthopedics in managing these conditions [26].

2. Diagnostic Analysis

Spectrum of Congenital Deformities

The spectrum of congenital deformities observed in this study was diverse, with clubfoot being the most prevalent, affecting 56 patients. This condition has been documented extensively in literature, with international studies indicating its occurrence in approximately 1 in 1000 live births [27]. **Dobbs et al. (2006)** noted that clubfoot is influenced by genetic factors and is often associated with familial incidence [27]. But in our study, we did not find any familial inheritance for the same.

There was an association seen with clubfoot AMC and CCBS. It was observed that 4 patients with clubfoot had a syndromic association with AMC. AMC is characterized by joint contractures affecting two or more areas of the body, frequently leading to non idiopathic clubfoot deformities that tend to be stiffer than those observed in idiopathic clubfoot cases. A study by **Singh et al. (2018)** in India indicated that patients with AMC often present with additional musculoskeletal abnormalities, complicating their treatment [28].

Also, there were 2 patients who had clubfoot in association with congenital constriction band syndrome. Clubfoot deformities have been reported to occur in a significant proportion of patients with CCBS. The affected foot may be situated below an ipsilateral band or may appear in a limb that lacks a proximal band. Researchers have observed that clubfeet associated with this syndrome are non idiopathic and tend to be rigid and resistant to nonoperative treatments, resulting in many patients from early studies requiring extensive surgical release.

The study identified other significant congenital defects, including radial club hand (7 patients) and tibia hemimelia (3 patients). The occurrence of radial club hand has been explored in multiple studies, where it was found to co-occur with syndromic conditions such as Holt-Oram syndrome. An international review by **Al-Qattan et al. (2013)** highlights that radial club hands can often be

part of a broader spectrum of congenital anomalies through their study on the genetic basis of failure of axis formation/differentiation of upper limb development [29].

The presence of syndromic conditions such as CHARGE syndrome (2 patients) and TORCH syndrome (1 patient) in patients with radial club hands were also noted. This indicates that radial club hand (RCH) can manifest in patients with heart defects and ear anomalies as part of a syndromic association. Furthermore, it emphasizes that children affected by TORCH infections, particularly those with congenital rubella syndrome, may exhibit immunodeficiency alongside limb defects. Notably, the child enrolled in this study had congenital rubella syndrome, Langerhans cell histiocytosis, and radial club hand. Research conducted by **Sadler et al**. highlights the necessity of thorough evaluations for children diagnosed with congenital limb defects to identify any potential associated syndromes [30] .

Syndactyly and Polydactyly were no less common defects noted during the study period. A total of 8 patients had syndactyly. In hand, mostly it was the middle and ring fingers which were found to be fused. Some patients with syndactyly of the hand were old stating the fact that it didn't affect their functional outcome much. A systematic review by **Hwang et al. (2011)** on the epidemiology of syndactyly indicated that it often occurs in conjunction with other congenital anomalies, reinforcing the need for thorough clinical assessments [31]. but we did not find any such correlation what we did find was synpolydactyly in 2 patients.

Polydactyly, was seen in 16 patients, is another very common congenital anomaly. It can variably have pre-axial or postaxial presentations including both hands and feet. An Indian study by **Choudhury et al. (2020)** reported that polydactyly often requires surgical intervention for

functional (patient might complain of pain or the extra digit getting stuck in things/clothing while working making it a problematic situation) and cosmetic reasons [32] .

We also observed patients with other conditions, including brachydactyly (4), macrodactyly (4)—one of whom was diagnosed with neurofibromatosis type 1—camptodactyly (2), clasp thumb (4), radio-ulnar synostosis (4), congenital vertical talus (3), and congenital pseudoarthrosis of the tibia (2). In the cases of congenital pseudoarthrosis, no history of neurofibromatosis type 1 was reported. Additionally, we identified CCBS as a distinct condition in 6 patients and arthrogryposis multiplex congenita (AMC) in 5 patients. Notably, many individuals with benign conditions such as brachydactyly sought hospital care for unrelated issues and were subsequently enrolled in the study as part of the CLD cohort; however, they reported no significant problems related to their existing defects.

In contrast, we also documented extremely rare conditions, such as two cases of proximal focal femoral deficiency: one involving a newborn and the other a 9-year-old male child. Both individuals exhibited additional bony deformities in the affected limb. Additionally, the newborn was noted to have a perimembranous ventricular septal defect (VSD). another rarer entity, Freeman Sheldon Syndrome (type of **congenital contracture syndrome)** was seen in one child with dysmorphic facies, bilateral clasp thumb and bilateral clubfeet. Freeman-Sheldon syndrome, also known as "whistling face syndrome," is a rare congenital disorder characterized by distinct

facial features, joint contractures, and limb abnormalities, particularly involving the hands and feet. This condition is most often caused by mutations in the **MYH3 gene**, which is involved in the development of skeletal muscle tissue [45]. The disorder is typically inherited in an **autosomal**

dominant manner, meaning affected individuals have a 50% chance of passing it on to their children [45]. Genetic counseling is essential to help families understand the risks associated with future pregnancies and the hereditary nature of the syndrome [46].

Also did we find patients with Larsen syndrome and congenital pterygium syndrome.

Larsen syndrome, a rare genetic condition triggered by **FLNB** gene mutations, causes severe joint dislocations, particularly in the knees, hips, and elbows, along with distinctive skeletal abnormalities. Beyond these, children with Larsen syndrome often present with noticeable craniofacial features like a flat midface and wide-set eyes, as well as congenital limb defects, including clubfoot, syndactyly (fused fingers or toes), or polydactyly (extra digits). To ensure comprehensive care, diagnostic tests like 2D Echo and abdominal ultrasound are crucial for detecting associated heart or organ issues, enabling timely intervention and preventing complications [48,49].

Congenital Pterygium Syndrome (CPS) is a rare genetic condition characterized by webs of skin (pterygia) that restrict movement, typically found at the joints (e.g., knees, elbows, neck). This syndrome can also present with various limb defects, most commonly **syndactyly** (webbed fingers or toes), **contractures** (permanent joint stiffness), and occasionally **clubfoot** or **limb hypoplasia** (underdeveloped limbs). The restricted joint mobility and limb malformations seen in CPS can severely impact physical function, often requiring surgical interventions to improve mobility and quality of life [50,51].

3. Limb Involvement Distribution

In terms of limb involvement, 71.7% of the patients had lower limb deformities, while 28.3% had upper limb deformities. This finding mirrors previous research, which consistently shows a higher prevalence of lower limb congenital anomalies [33][34] . A recent Indian study by **Jain et al. (2021)** found that lower limb defects, particularly clubfoot, constituted the majority of congenital limb deformities, necessitating targeted intervention strategies [35] . Their results revealed that 78% of the congenital limb defects affected the lower limbs out of a total 150 patients , indicating

a significant predisposition for lower limb involvement. Conditions such as clubfoot and congenital vertical talus were among the most frequently diagnosed anomalies. And this result was corroborative with our study where we also found around 72% affection of the lower limbs. In another study conducted by **Bianchi, F. et al. (2016),** their study reported that of the 120 patients diagnosed with congenital limb deformities, 72% exhibited lower limb involvement

4. Key Associations with Syndromic Conditions

The study highlights several deformities with significant syndromic associations. Notably, AMC and CCBS were observed in conjunction with clubfoot, aligning with findings from a cohort study by **Gupta et al. (2018)**, which indicated that about 30% of patients with clubfoot had additional syndromic features [36] . Many other deformities like RCH, Macrodactyly, PFFD etc showed syndromic associations as well.

Eight patients with congenital conditions like —ranging from radial club hand, congenital rubella syndrome, polydactyly, PFFD, and more—underwent investigations like 2D Echo and abdominal ultrasounds to uncover underlying health issues if any.

- **CHARGE Syndrome**: Both heart and abdominal scans came back normal.
- **Polydactyly with Scoliosis**: in the child a **2 mm PDA** was detected (no active intervention done as it was thought to resolve with time), while the abdominal ultrasound was clear.
- **PFFD**: Diagnosed with a perimembranous VSD in 2D ECHO and undescended testes in USG-W/A based on the scans.
- **Congenital Rubella Syndrome**: Eye and ear exams were normal, but signs of **premature brain parenchyma** were likely tied to early birth.
- **Pterygium Syndrome & Polydactyly**: No further defects discovered. Both 2D ECHO and USG W/A were normal.

STRENGTHS AND LIMITATIONS

- **Strengths of the Study**
- **Comprehensive Analysis**: The study includes a diverse patient population of 121 individuals seen over a period of 18 months at a tertiary institution with various congenital deformities, offering valuable insights into the prevalence of conditions such as clubfoot and polydactyly. This enhances the findings' generalizability.
- **Demographic Insights**: It effectively highlights key demographic factors, notably the higher incidence of deformities in males and a predominance in pediatric patients. These findings align with global trends and inform future screening and treatment strategies.
- **Syndromic Associations**: The identification of syndromic associations, such as AMC and CHARGE syndrome, underscores the need for interdisciplinary care and genetic screening for patients with suspected syndromes.
- **Focus on Limb Involvement**: By distinguishing between upper and lower limb deformities, the study emphasizes the higher frequency of lower limb issues, which is crucial for prioritizing interventions aimed at improving mobility in affected patients.
- **Limitations of the Study**
- **Lack of Long-term Follow-up**: The study does not include long-term assessments of patient outcomes post-treatment, limiting insights into treatment efficacy and quality of life.
- **Limited Functional Assessments**: There is no evaluation of the functional impact of deformities on patients' daily lives, missing an opportunity to gather data on mobility and psychosocial well-being.

- **No Risk Factor Analysis**: The study does not investigate genetic or environmental factors contributing to congenital deformities, which could inform preventative strategies.
- **Single-Center Study**: Conducted in a single healthcare center, the findings may not be generalizable to different regions or populations, highlighting the need for multicenter studies.
- **Absence of Treatment Comparisons**: The study does not compare outcomes of various treatment modalities, making it difficult to determine the optimal approach for managing deformities.
- **Syndromic Diagnosis Without Genetic Confirmation**: Diagnoses of complex syndromic conditions were likely made without genetic testing, which could lead to misclassification. Future research should incorporate genetic confirmation.
- **Focus on Diagnosis Over Treatment Efficacy**: The study emphasizes the prevalence and types of deformities but lacks information on treatment outcomes, which is essential for guiding clinical practice.

SUMMARY

This study investigates the distribution and characteristics of congenital limb defects (CLDs) among patients at a tertiary healthcare center. Congenital limb defects encompass a wide range of structural anomalies present from birth, which can significantly impact both physical functionality and psychological well-being. These defects result from disruptions during intrauterine development and may be associated with genetic, environmental, or idiopathic factors. Despite the burden these conditions impose on individuals and healthcare systems, research on CLDs, particularly in India, remains scarce. This study aims to fill that gap by providing valuable insights into the types, prevalence, and associations of CLDs in patients treated at a tertiary care hospital in central India.

The primary objective of this hospital-based observational study was to assess the distribution of different congenital limb defects among patients attending the Orthopedics, Pediatrics, and Burns and Plastic Surgery departments at AIIMS Bhopal from March 2023 to September 2024. The study enrolled 121 patients diagnosed with congenital limb defects, with data collected through detailed clinical evaluations, including patient history, physical examination, radiographic imaging, and genetic testing when necessary. The secondary objectives aimed to explore the health-seeking behavior of patients and their families regarding congenital limb defects and to assess the quality of life of the affected individuals using patient or parent-reported outcome measures. However, except for an analysis of out-of-pocket expenditure on treatment, the other secondary objectives were not fulfilled.

The demographic analysis of the study cohort revealed a significant gender disparity, with 67.5% of the patients being male and 32.5% female. This gender disparity is consistent with similar findings from other studies, suggesting that males may have a higher genetic or environmental predisposition to congenital limb defects. Most patients (85%) were under the age of 18, highlighting that these defects are primarily diagnosed and treated in childhood. This underscores the importance of early detection and intervention in managing congenital limb deformities, as early treatment often leads to better functional outcomes and less invasive corrective procedures.

The study identified a wide spectrum of congenital limb defects among the participants, with clubfoot emerging as the most common condition, affecting 56 patients. Clubfoot was observed in both unilateral and bilateral presentations, and in some cases, it was associated with syndromic conditions like arthrogryposis multiplex congenita (AMC) and congenital constriction band syndrome (CCBS). These associations suggest that non-idiopathic forms of clubfoot, particularly those linked with syndromes, may be more complex and resistant to standard non-operative treatments. Polydactyly and syndactyly were also frequently encountered, affecting 16 and 8 patients, respectively. Polydactyly manifested in both pre-axial and post-axial forms, affecting hands and feet, while syndactyly typically involved fusion between the middle and ring fingers. Both conditions are well-documented congenital anomalies, often requiring surgical correction for functional and cosmetic reasons. Radial club hand, seen in 7 patients, was often associated with syndromic conditions such as CHARGE syndrome and Holt-Oram syndrome, indicating the complex interplay of genetic factors in these malformations.

An important finding of the study was the high prevalence of syndromic associations in patients with congenital limb defects. In addition to CHARGE and Holt-Oram syndromes, the study also found that TORCH infection was implicated in case of RCH, further highlighting the role of infections during pregnancy in the development of certain congenital limb defects.The study also identified rarer conditions such as tibia hemimelia, congenital vertical talus, camptodactyly, and congenital pseudoarthrosis of the tibia, emphasizing the diverse spectrum of congenital limb deformities managed at the institution. These rarer conditions present unique challenges in terms of management, often requiring specialized surgical interventions and long-term follow-up care. The study also emphasized the critical role of diagnostic investigations, particularly 2D echocardiography and ultrasound of the abdomen and pelvis (USG W/A) in assessing patients with congenital limb defects.

Although the study intended to assess quality of life and patient-reported outcomes using standardized scoring systems, these secondary objectives were not fulfilled. The only secondary objective that was addressed in detail was the analysis of out-of-pocket expenditure incurred by families for the treatment of congenital limb defects. However, the out-of-pocket expenditure assessment was largely subjective and did not provide comprehensive insights into the true financial burden on families. It was not of much help in the study as it failed to capture the full impact of the costs on the families' financial situations, limiting its usefulness in drawing broader conclusions about the economic challenges faced by those undergoing treatment for congenital limb defects.

In conclusion, this study offers valuable insights into the spectrum of congenital limb defects in a tertiary care setting in India. It emphasizes the importance of early diagnosis, multidisciplinary management, and affordable healthcare access for individuals with

congenital limb defects. The high prevalence of syndromic associations highlights the need for comprehensive evaluations and genetic testing in these patients to guide appropriate treatment strategies. Furthermore, the study calls for increased public awareness to encourage early health-seeking behavior and reduce the stigma associated with congenital deformities. By providing insights into the prevalence and distribution of congenital limb defects, this research aids in improving healthcare planning and optimizing resource allocation for the management of these conditions in developing countries, like ours.

CONCLUSION

In conclusion, this study successfully achieved its primary aim of mapping the distribution of congenital limb defects, shedding light on the prevalence and variety of these conditions. While the secondary goals—exploring health-seeking behaviors and quality of life—were not accomplished, the research offers valuable insights into the importance of early diagnosis and holistic care. These findings highlight the need for timely intervention and multidisciplinary management to improve outcomes for individuals affected by congenital limb anomalies.

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ANNEXURES

ANNEXURE 1

PROFORMA

- PATIENT HISTORY:
    - Name
    - Age
    - Age of the parents at the time of conception
    - Sex
    - Address / Contact details
    - Chief Complaints
    - Birth history, if known
    - Family history
    - Treatment history
    - Pedigree chart
    - Consanguinity
    - Gestational age at which the child is born, if known
    - Risk factors – drugs/ smoking/ alcohol/ TORCH any other significant risk history if details are available
    - Additional remarks from the patient and attendant

- GENERAL EXAMINATION:

- LOCAL EXAMINATION:

    - Type of anomaly-

    - Inspection

    - Palpation

    - Measurements

- **INVESTIGATIONS:** (done as part of routine work up of patient)

1. Radiographs of the affected part in two orthogonal views preferably AP, Lateral.

2. Genetic screening, if done

3. Haematological markers

4. 2D ECHO, USG abdomen, USG KUB, Cystourethrogram, barium studies.

ANNEXURE 2

Participant Information Sheet

STUDY TITLE: To assess the spectrum of patients with congenital limb defect s resenting at a tertiary care hospital**.**

1. STUDY INVESTIGATORS:

Student: Dr, Priya Maheshwari, Junior Resident, Department of Orthopaedics **Guide**: Dr John A Santoshi, Professor, Department of Orthopaedics**Co-guides:** Dr. Shikha Malik, Professor, Department of Paediatrics

Dr. Manal Mohd. Khan, Professor, Department of Burns & Plastic Surgery Dr. Prateek Behera, Additional Professor, Department of Orthopaedics

2. Introduction:

We are conducting research to assess the spectrum of patients with congenital limb defects presenting at a tertiary care hospital. If you agree to take part voluntarily, we will ask you to sign a consent form.

3. Purpose of the study:

The aim of our study is to assess the spectrum of patients with congenital limb defects presenting at a

tertiary care hospital through which the spectrum and distribution of the same will be assessed and recorded. This would help generate data to help the treating

physicians as well as policy makers for effective planning by doing the cost analysis of thesurgeries if the patient undergoes corrective procedures and allocation of resources/funds for the management of individuals with congenital limb defects and also to understand thehealth seeking behaviour by asking the patient/parent about their attitude towards the defect and have they ever considered about undergoing any medical procedure for the eradicationof the same.

4. Taking part – what it involves:

a. Do I have to take part?

Participation in this research is entirely voluntary. A decision to not take part, or towithdraw at any time, will not affect your rights or medical care in any way.

b. What is expected of me if I take part?

If you participate, we will take your medical history. Clinical photographs/ imagesof the patient and/or involved body part(s) would be taken. There is any chance that you may be identifiable from a photograph or other static or moving image, or fromits legend or accompanying text in the published thesis or publication in a scientificjournal. Consent for recording and publishing your identifiable information is expected.

c. How long will my part in the study last?

Your participation in the study would be one time.

d. Do I have to come for extra visits or undergo extra investigations for the study?

No extra visits needed.

e. What are the possible benefits in taking part

Your treatment would continue as per your discussion with the treating doctor. There would be no specific benefits to you in taking part in this study.

f. What are the possible disadvantages and risks of taking part?

There are no foreseeable disadvantages or risks because of the study. There may be risk of loss of privacy and confidentiality of information collected even thoughall possible measures will be taken to ensure safe data storage and access.

g. What happens to the information collected from me?

All information collected will be kept strictly confidential. Results from the studywill be reported as group data and will not identify you in any way. However, youmay be identifiable from a photograph or other static or moving image, or from itslegend or accompanying text in the published thesis or publication in a scientific journal.

h. What if I change my mind regarding participation in the study?

You are entitled to change your mind and withdraw at any time during the studywithout any disadvantage or penalty.

i. Whom do I contact for more information or if I have further questions or concerns?

If you want any information or have questions or concerns, you can contact:

Dr. Priya Maheshwari

Post graduate student,

Department of Orthopaedics,

AIIMS, Bhopal

8827166546

priyamaheshwari094@gmail.com

Dr. Shashank Purwar

Member Secretary,

Institute Ethics Committee – Student Research7773010099

ihecpgr@aiimsbhopal.edu.in

ANNEXURE 3

प्रतिभागी सूचना पत्र

1. **अध्ययन शीर्षकः** तृतीयक देखभाल अस्पताल में उपस्थित जन्मजात अंग दोष वाले रोगियों के स्पेक्ट्रम का आकलन करना।

   अध्ययन अन्वेषकः छात्रः डॉ. प्रिया माहेश्वरी, जूनियर रेजिडेंट, ऑर्थोपेडिक्स विभाग

   मार्गदर्शकः डॉ. जॉन ए संतोषी, प्रोफेसर, ऑर्थोपेडिक्स विभाग

   सह–मार्गदर्शकः डॉ. शिखा मलिक, प्रोफेसर, बाल रोग विभाग, डॉ. मनाल मोहम्मद खान, प्रोफेसर, बर्न्स और प्लास्टिक सर्जरी विभाग, डॉ. प्रतीक बेहरा, अतिरिक्त प्रोफेसर, ऑर्थोपेडिक्स विभाग

2. **परिचयः** हम तृतीयक देखभाल अस्पताल में उपस्थित जन्मजात अंग दोष वाले रोगियों के स्पेक्ट्रम का आकलन करने के लिए अनुसंधान कर रहे हैं। यदि आप स्वेच्छा से भाग लेने के लिए सहमत हैं, तो हम आपसे सहमति पत्र पर हस्ताक्षर करने के लिए कहेंगे।

3. **अध्ययन का उद्देश्यः** हमारे अध्ययन का उद्देश्य तृतीयक देखभाल अस्पताल में उपस्थित जन्मजात अंग दोष वाले रोगियों के स्पेक्ट्रम का आकलन करना है, जिसके माध्यम से उनके स्पेक्ट्रम और वितरण का आकलन और

रिकॉर्ड किया जाएगा। इससे उपचार करने वाले चिकित्सकों के साथ–साथ नीति निर्माताओं को सर्जरी की लागत का विश्लेषण करके प्रभावी योजना बनाने में मदद मिलेगी, यदि रोगी सुधारात्मक प्रक्रियाओं से गुजरता है और जन्मजात अंग दोष वाले व्यक्तियों के प्रबंधन के लिए संसाधनों/धन का आवंटन करता है और रोगी/माता–पिता से दोष के प्रति उनके दृष्टिकोण के बारे में पूछकर स्वास्थ्य चाहने वाले व्यवहार को समझने में मदद मिलेगी और क्या उन्होंने कभी इसके उन्मूलन के लिए किसी चिकित्सा प्रक्रिया से गुजरने के बारे में सोचा है।

4. भाग लेना – इसमें क्या शामिल है:. क्या मुझे भाग लेना है? इस शोध में भागीदारी पूरी तरह से स्वैच्छिक है। भाग न लेने या किसी भी समय वापस लेने का निर्णय किसी भी तरह से आपके अधिकारों या चिकित्सा देखभाल को प्रभावित नहीं करेगा।

(क) अगर मैं भाग लेता हूं तो मुझसे क्या अपेक्षा की जाती है?

अगर आप भाग लेते हैं, तो हम आपका मेडिकल इतिहास लेंगे। रोगी और/या प्रभावित शरीर के अंग(अंगों) की नैदानिक तस्वीरें/छवियाँ ली जाएँगी। इस बात की कोई संभावना है कि आप किसी तस्वीर या अन्य स्थिर या गतिशील छवि से, या प्रकाशित थीसिस या वैज्ञानिक पत्रिका

में प्रकाशन में इसके लेजेंड या साथ में दिए गए पाठ से पहचाने जा सकते हैं। आपकी पहचान योग्य जानकारी को रिकॉर्ड करने और प्रकाशित करने के लिए सहमति अपेक्षित है।

(ख) अध्ययन में मेरा हिस्सा कितने समय तक चलेगा?

अध्ययन में आपकी भागीदारी एक बार की होगी।

(ग) क्या मुझे अध्ययन के लिए अतिरिक्त विज़िट के लिए आना होगा या अतिरिक्त जाँच करवानी होगी?

अतिरिक्त विज़िट की आवश्यकता नहीं है।

(घ) भाग लेने से संभावित लाभ क्या हैं

आपका उपचार उपचार करने वाले डॉक्टर के साथ आपकी चर्चा के अनुसार जारी रहेगा। इस अध्ययन में भाग लेने से आपको कोई विशेष लाभ नहीं होगा।

(ड) भाग लेने के संभावित नुकसान और जोखिम क्या हैं?

अध्ययन के कारण कोई संभावित नुकसान या जोखिम नहीं है। एकत्रित की गई जानकारी की गोपनीयता और गोपनीयता के नुकसान का जोखिम हो सकता है, भले ही सुरक्षित डेटा भंडारण और पहुँच सुनिश्चित करने के लिए सभी संभव उपाय किए जाएँगे।

(च.) मुझसे एकत्रित की गई जानकारी का क्या होगा?

एकत्र की गई सभी जानकारी को पूरी तरह से गोपनीय रखा जाएगा। अध्ययन के परिणामों को समूह डेटा के रूप में रिपोर्ट किया जाएगा और किसी भी तरह से आपकी पहचान नहीं की जाएगी। हालाँकि, आप किसी फ़ोटो या अन्य स्थिर या चलती छवि से, या प्रकाशित थीसिस या वैज्ञानिक पत्रिका में प्रकाशन में इसके लेजेंड या साथ के पाठ से पहचाने जा सकते हैं।

(छ) अगर मैं अध्ययन में भागीदारी के बारे में अपना विचार बदल देता हूँ तो क्या होगा?

आप बिना किसी नुकसान या दंड के अध्ययन के दौरान किसी भी समय अपना विचार बदलने और वापस लेने के हकदार हैं।

(ज) अधिक जानकारी के लिए या अगर मेरे पास और प्रश्न या चिंताएँ हैं तो मैं किससे संपर्क करूँ? यदि आपको कोई जानकारी चाहिए या आपके कोई प्रश्न या चिंताएँ हैं, तो आप संपर्क कर सकते हैं:

डॉ. प्रिया माहेश्वरी
स्नातकोत्तर छात्र,
ऑर्थोपेडिक्स विभाग,
एम्स, भोपाल,
मो. नं. – 8827166546
ई–मेल आई.डी. – priyamaheshwari094@gmail.com

डॉ. शशांक पुरवार
सदस्य सचिव,
संस्थान आचार समिति– छात्र अनुसंधान
मो. नं. – 7773010099
ई–मेल आई.डी. – ihecpgr@aiimsbhopal.edu.in

ANNEXURE 4

प्रतिभागी सूचना सहमति प्रपत्र

[PARTICIPANT INFORMED CONSENT FORM (PICF)]

अध्ययन के लिए प्रतिभागी का पहचान नंबर / Participant Identification Number for Study: ……..

अनुसंधान का शीर्षक / Title of Project :Spectrum of Patients with Congenital Limb Defects Presenting at a Tertiary Care Hospital

.

छात्र शोधकर्ता का नाम / Name of Student Investigator : Dr Priya Maheshwari

शोध मार्गदर्शक का नाम / Name of Thesis Guide : Dr John A Santoshi

मैंने संस्करण संख्या ॥ ३३ दिनांक 15–01–2023 के सूचना पत्रक में दिए गए सभी तथ्यों को सावधानीपूर्वक पढ़ लिया है / मुझे अपनी भाषा में स्पष्ट रूप से विस्तारपूर्वक समझा दिया गया है, और मैंने तथ्यों को पूर्णतः समझ लिया है। मैं पुष्टि करता / करती हूँ, कि मुझे सवाल पूछने के अवसर प्रदान किए गए।

The contents of the information sheet version II dated 15th January 2023 that was provided have been read carefully by me / explained in detail to me, in a language that I comprehend, and I have fully understood the contents. I confirm that I have had the opportunity to ask questions.

मुझे इस अध्ययन की प्रकृति एवं उद्देश्य और इसके संभावित जोखिमों/लाभों तथा अध्ययन की अनुमानित अवधि एवं अन्य प्रासंगिक जानकारी के बारे में विस्तारपूर्वक समझा दिया गया है। मुझे पता है कि इस अध्ययन में मेरी भागीदारी स्वैच्छिक है तथा मैं इस अध्ययन से किसी भी समय बिना कोई कारण बताए, अपना नाम वापस लेने के लिए स्वतंत्र हूं और नाम वापस लेने पर मेरी चिकित्सा देखभाल या कानूनी अधिकारों पर कोई प्रभाव नहीं पड़ेगा।

The nature and purpose of the study and its potential risks/benefits and expected duration of the study, and other relevant details of the study have been explained to me in detail. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal right being affected.

मैं समझता/समझती हूं कि इस अनुसंधान में मेरी भागीदारी से मेरे बारे में एकत्र की गई जानकारी और मेरे चिकित्सीय दस्तावेजों के किसी भी भाग को एम्स के जिम्मेदार व्यक्तियों द्वारा ही देखा जाएगा। मैं इन व्यक्तियों को मेरे रिकॉर्ड प्राप्त करने की अनुमति प्रदान करता/ करती हूँ।

I understand that the information collected about me from my participation in this research and sections of any of my medical notes may be looked at by responsible individuals from AIIMS. I give permission for these individuals to have access to my records.

यदि प्रतिभागी बच्चा है, तो उसकी मौखिक या लिखित सहमति प्राप्त की गई है।

If participant is child, his / her verbal or written assent has been obtained.

मैं उक्त अध्ययन में भाग लेने के लिए सहमत हूं।

I agree to take part in the above study.

_____

(प्रतिभागी के हस्ताक्षर / बांए अंगूठे का निशान)

(Signature / Left Thumb Impression)

दिनांक/Date .....

स्थान/Place .....

प्रतिभागी का / Name of the participant .....

पिता/पति/पत्नी का नाम /Son/Daughter/Spouse of.....

डाक का पूरा पता /Complete Postal Address .....

अगर अशिक्षित हो / If illiterate,

सूचना पत्र को प्रतिभागी को पढ़ा और समझाया गया है, और प्रश्न पूछने का अवसर दिया गया है। मैं पुष्टि करता हूं कि व्यक्ति ने स्वतंत्र रूप से सहमति दी है।

The information sheet has been read and explained to the participant, and given the opportunity to ask questions. I confirm that the individual has given consent freely.

कानूनी रूप से स्वीकार्य प्रतिनिधि अथवा निष्पक्ष गवाह के हस्ताक्षर

Signature of Legally Acceptable Representative or impartial witness: …..……

दिनांक / Date: .....

एलएआर अथवा निष्पक्ष गवाह का नाम /

Name of LAR or impartial witness: /.....

पता / Address : .....

प्रमाणित किया जाता है कि उक्त सहमति मेरी उपस्थिति में प्राप्त की गई ।

This is to certify that the above consent has been obtained in my presence.

_____

छात्र शोधकर्ता के हस्ताक्षर दिनांक.....

स्थान..... Date ……

Signature of the Student Investigator Place

……

कृपया ध्यान दें: सहमति प्रपत्र तीन प्रतियों में (1) रोगी के लिए, (2) शोधकर्ता, (3) संस्थान के लिए तैयार करें।

Please note: Three copies should be made, for (1) Patient, (2) Researcher, (3) Institution

ANNEXURE 5

Parental Consent Form

I,_____Father / Mother of_____am the lawful guardian of childhaving_____disease/defect. The medical condition of my child has beenexplained to me very well in my own language therefore I'm giving my consent for the participation of my child in the research.

By this I grant permission to take medical history, clinical photography/images of the child and/or involved body part(s). You can use above identifiable data containing static or movingimages of my child in the published thesis or publication in a scientific journal.

Participation in this research will be entirely voluntary and I can revoke or withdraw participation of my child at any time without affecting my rights or medical care in any way.

Name……

Place…….

Phone no……..

Left Thumb Impression

Signature……

Date ….. /….. /……

ANNEXURE-6

माता-पिता की सहमति प्रपत्र

मैं, पिता/माता, रोग/दोष से ग्रस्त बच्चे का वैध अभिभावक हूँ। मेरे बच्चे की चिकित्सा स्थिति मुझे मेरी अपनी भाषा में बहुत अच्छी तरह से समझाई गई है, इसलिए मैं अपने बच्चे को शोध में भाग लेने के लिए अपनी सहमति दे रहा हूँ।

इसके द्वारा मैं बच्चे का चिकित्सा इतिहास, नैदानिक फोटोग्राफी/चित्र और/या शामिल शरीर के अंग(अंगों) को लेने की अनुमति देता हूँ। आप मेरे बच्चे की स्थिर या चलती छवियों वाले उपरोक्त पहचान योग्य डेटा का उपयोग प्रकाशित थीसिस या वैज्ञानिक पत्रिका में प्रकाशन में कर सकते हैं।

इस शोध में भागीदारी पूरी तरह से स्वैच्छिक होगी और मैं किसी भी तरह से अपने अधिकारों या चिकित्सा देखभाल को प्रभावित किए बिना किसी भी समय अपने बच्चे की भागीदारी को रद्द या वापस ले सकता हूँ।

नाम :– .....

स्थान :–.....

फ़ोन नंबर :–..... बायाँ अंगूठा छाप

हस्ताक्षर .....

दिनांक .....

ANNEXURE 7

Verbal Assent Form

Thesis Title: To assess the spectrum of patients with congenital limb defects presenting at a tertiary care hospital.

Investigator: Dr. Priya Maheshwari

Hello, I am Dr Priya Maheshwari and I am conducting a thesis on "*To assess the spectrum of patients with congenital limb defects presenting at a tertiary care hospital*".

If you agree to be in our research, we will ask you certain set of questions regarding the medical history and clinical photography/images of the child involving body parts will be taken.

You are allowed to ask question at any time and can also stop or skip any question while conducting the study.

Do you understand what I am saying and are you willing to?

TO BE COMPLETED BY PERSON OBTAINING VERBAL ASSENT FROM THE CHILD:

Child's Response : Yes ☐ No ☐

ANNEXURE 8

Master sheet of data collection

	AGE	SEX	DIAGNOSIS	INSPECTION	SIDE	OTHER ASSOCIATED FEATURES	INVESTIGATIONS- 2D echo	BIRTH HISTORY
case 1		2 F	Tibia Hemimelia with lateral subluxation of ankle joint	lower limb length discrepancy, with lateral subluxation of ankle joint	Right	NA	Not done	Normal vaginal delivery
case 2	5 months	F	Bilateral equinocanovarous deformity with ARC	bilateral equinocanovarous deformity with adduction, internally rotated and pronated upper limb	right and left	NA	Not done	normal vaginal delivery
case 3		2 M	radial club hand	hand was bent radially with small underdeveloped toe on the same side and cleft palate with defect in both eyes	right	Microbia, cleft palate, coloboma of eye	2D echo- WNL USG WIA-WNL	Normal vaginal delivery
case 4	15 days	M	pre axial polydactyly of foot	rudimentary thumb present	Left	NA	Not done	LSCS
case 5		2.5 F	equinocavovarus deformity	equinocavovarus deformity of left foot	left	NA	Not done	Normal Vaginal Delivery
case 6		7 M	Congenital vertical talus	rocker bottom foot	Right	NA	Not done	normal vaginal delivery
case 7		1.5 F	equinocavovarus deformity	equinocavovarus deformity	right	NA	Not done	Normal vaginal delivery
case 8		1.5 M	equinocavovarus deformity	equinocavovarus deformity	right	NA	Not done	Normal Vaginal Delivery
case 9		9 M	Syndactyly of thumb and index finger	fused thumb and index finger	left	none	Not done	LSCS
case 10		1 F	Preaxial polydactyly right hand, syndactyly of 2nd web space, syndactyly 1st web space left	Preaxial polydactyly right hand, syndactyly of 2nd web space, syndactyly 1st web space left	right & left	none	Not done	Normal vaginal delivery
case 11		15 F	preaxial polydactyly right thumb	rudimentary thumb present	right	none	Not done	Normal vaginal delivery
case 12		4 F	bilateral equinocanovarous deformity	equinocavovarus deformity	right &left	none	Not done	Normal vaginal delivery
case 13		1 M	congenital constrictive band syndrome with bilateral equinocavovarous deformity	equinocavovarous deformity with absent digits	right and left	none	Not done	Normal vaginal delivery
case 14		12 M	congenital patella dislocation	genu valgum deformity with lateralized patella bilaterally	right and left	stunted growth with macrotia with ocassi	Not done	Normal vaginal delivery
case 15		8 M	Arthrogryposis of right hand with clamp thumb	adducted thumb present with fibrous tissue seen as cord like structure	right	none	Not done	normal vaginal delivery
case 16		10 M	flexion contracture of bilateral little fingers	little fingers were contracted, passive extension was possible	right & left	none	Not done	Normal vaginal delivery
case 17		15 M	post-axial polydactyly of little of left foot	rudimentary toe lateral side of 5th toe	left	none	Not done	Normal vaginal delivery
case 18		1 M	equinocavovarus deformity	equinocavovarus deformity of bilateral feet	right & left	none	Not done	Normal vaginal delivery
case 19		1 M	equinocavovarus deformity with congenital constriction band syndrome	bilateral equinocanus deformity with loss of digits in feet	right & left	none	Not done	Normal vaginal delivery
case 20		30 M	congenital constriction band syndrome	all 4 limbs had loss of digits	all 4 limbs	none	Not done	Normal vaginal delivery
case 21		24 F	thumb hypoplasia	left thumb was hypoplastic with rudimentary adduction muscle	left	none	Not done	normal vaginal delivery
case 22		49 F	Macrodactyly with lipofibromatosis of index finger	index finger was bigger and was painful than other fingers	right	none	Not done	Normal vaginal delivery
case 23		64 F	Syndactyly of middle and ring finger	middle and ring finger fused throughout length	left	none	Not done	Normal vaginal delivery
case 24		30 F	preaxial polydactyly of right thumb	extra thumb present on the radial aspect of normal thumb- Wassel Type 7	right	none	Not done	Normal vaginal delivery
case 25		65 F	brachydactyly of 4th toes of bilateral feet	brachydactyly of 4th toes of bilateral feet	right and left	none	Not done	Normal vaginal delivery
case 26		1 F	Syndactyly of middle and ring finger	middle and ring finger fused throughout length	left	none	Not done	LSCS
case 27		4 F	congenital pseudarthrosis of tibia	anterolateral bowing of leg present	left	none	Not done	normal vaginal delivery
case 28		3 M	equinocavovarus deformity	Equinocavovarus deformity of right foot	right	none	Not done	normal vaginal delivery
case 29	5months	M	equinocavovarus deformity	Equinocavovarus deformity of right and left foot	right & left	none	Not done	Normal vaginal delivery
case 30	2 months	M	equinocavovarus deformity	Equinocavovarus deformity of right and left foot	right & left	none	Not done	Normal vaginal delivery
case 31		2 M	postaxial polydactyly right hand	extra hypoplastic thumb present radially over right hand	right	none	Not done	normal vaginal delivery
case 32		53 M	congenital constriction band syndrome	all 4 fingers absent in left hand with rudimentary thumb	Left	none	Not done	normal vaginal delivery
case 33		16 M	postaxial polydactyly in bilateral lower limb with simple syndactyly in bilateral upper limb	left hand- syndactyly present IMK digits and right- MK digits with preaxial polydactyly, feet- polyaxial polydactyly	right & left	none	Not done	normal vaginal delivery
case 34		80 M	congenital constriction band syndrome	all 4 fingers absent in right hand with rudimentary thumb	right	none	Not done	Normal vaginal delivery
case 35		9 M	congenital ulnar deficiency	cubitus varus deformity	right	none	Not done	LSCS
case 36		17 M	neurofibromatosis type 1 with macrodactyly	macrodactyly of right ring finger	right	cafe au lait spots with multiple neurofibr	not done	normal vaginal delivery
case 37		5 M	equinocavovarus deformity	Equinocavovarus deformity of left foot	left	none	Not done	LSCS
case 38		3 M	equinocavovarus deformity	Equinocavovarus deformity of right foot	right	none	not done	normal vaginal delivery
case 39		9 M	equinocavovarus deformity	Equinocavovarus deformity of left foot	left	none	Not done	normal vaginal delivery
case 40		31 M	syndactyly of MRC fingers of bilateral hands	fused middle, ring and little fingers of bilateral hands	right and left	none	not done	normal vaginal delivery
case 41		6 M	brachysyndactyly- short hand	all five fingers of left hand were fused with rudimentary thumb	left	Poland syndrome with absent chest wall	not done	normal vaginal delivery
case 42		15 F	bilateral radial club hands	radial club hand bilaterally with absent fingers	right and left	bronchiectasis with bilateral club hands	not done	normal vaginal delivery
case 43		16 M	macrodactyly of 2nd toe	macrodactyly of 2nd toe-left	left	none	not done	normal vaginal delivery
case 44		18 M	syndactyly of radius and ulna	valgus deformity over right elbow	right	none	not done	normal vaginal delivery

CONSANGUINITY	FAMILY HISTORY	TREATMENT HISTORY	SURGICAL/NON-SURGICAL	Procedure performed	IMPACT ON LIFE	OUT OF POCKET EXPENDITURE	GESTATIONAL AGE	RISK FACTORS- Drugs, Smoking, Alcohol, TORCH, Ot
no	NA	Yes	Surgical	resection of pseudoarthrosis and fixation with intramedullary nail	cosmesis and inability to bear weight on right lower limb	ayushman	Term delivery	none
No	NA	Yes	Surgical	serial casting- posteromedial soft tissue release	cosmesis and future difficulties in walking	Ayushman	Term delivery	none
No	NA	Yes	Surgical	centralization of right ulnar with soft tissue release	cosmesis and future difficulties in using right hand	BPL	Term delivery	none
No	NA	No			cosmesis	NA	Term delivery	none
no	NA	Yes	non Surgical	serial casting- Ponseti's method	cosmesis and future difficulties in walking	APL-20000	Term delivery	none
no	NA	No			cosmesis and future difficulties in walking	NA	Term delivery	none
no	NA	yes	non Surgical	serial casting- Ponseti's method	cosmesis and future difficulties in walking	APL-20000	Term delivery	none
no	NA	Yes	non surgical	serial plaster- 5 months- FAB	cosmesis and future difficulties in walking	APL- 15000	Term delivery	None
No	NA	no					Term delivery	none
no	NA	yes	surgical	syndactyly release	cosmesis and future difficulties in using right hand	APL-15000 INR	Term delivery	none
no	NA	yes	surgical	excision of rudimentary thumb	pain and thumb getting stuck in things/clothing affecting functionality	Ayushman	Term delivery	none
no	NA	yes	non surgical	serial plaster application	cosmesis and future difficulties in walking	APL-5000	Term delivery	none
no	NA	yes	non Surgical	serial plaster application	cosmesis and future difficulties in walking	APL-5000 INR	Term delivery	none
no	NA	yes	surgical	soft tissue release with medial plication	mainly to run or walk fast	Ayushman	Term delivery	none
no	NA	no					Term delivery	none
no	NA	no				BIA	Term delivery	none
no	NA	no					Term delivery	none
no	NA	yes	no surgical	serial plaster application	cosmesis and future difficulties in walking	APL-10000	Term delivery	none
no	NA	yes	non Surgical	serial plaster application	cosmesis and future difficulties in walking	APL- 15000	Term delivery	none
no	NA	No			no impact as such	BIA	Term delivery	none
no	NA	No			limited grasping of hand	BIA	Term delivery	none
no	NA	started taking treatment- planned for surgical management		yet to be performed	painful finger movement - difficulty in using the hand	BPL	Term delivery	none
no	NA	No				BIA	Term delivery	none
no	NA	No				BIA	Term delivery	none
no	NA	no				BIA	Term delivery	none
no	NA	yes	Surgical	syndactyly release	patient was young- decision was taken by family as the patient was un	APL- 10000 INR	Term delivery	none
no	NA	yes	Surgical	resection of pathological segment with Rush rod+ fibrare + bone grafting	limb length discrepancy- difficulty in walking	Ayushman	Term delivery	none
no	NA	yes	Non surgical FIB surgical	serial casting for 6 months followed by tenotomy	difficulty in walking	APL-70000 INR	Term delivery	none
no	NA	yes	Non surgical	foot abduction brace- 23 hours/day- to continue FAB for 2 months	cosmesis and future difficulties in walking	APL-3000	Term Delivery	none
no	NA	yes	non surgical	serial casting using ponseti method	cosmesis and future difficulties in walking	APL- 3000	Term delivery	none
no	NA	yes	surgical	excision of rudimentary thumb	cosmesis	APL-2000	term delivery	none
no	NA	no				BIA	term delivery	none
no	NA	no				BIA- if planning for surgery then Ayus	term delivery	none
no	NA	no					term delivery	none
no	NA	no					term delivery	none
no	grand mother had lyos	surgical	debulking of the finger		heaviness in finger- made it difficult to hold objects, while	ayushman	term delivery	none
no	NA	yes	surgical	tenotomy	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none
no	NA	yes	surgical	tenotomy 1b TA tendon transfer	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none
no	na	yes	surgical	tenotomy 1b tibialis anterior transfer and TA lengthening	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none
no	NA	no					term delivery	none
no	NA	no					term delivery	none
no	NA	for bronchiectasis- short medicinal		admitted in pediatrics ward for management of bronchiectasis	patient is intellectually disabled with functional compromise of bilateral hands		pre-term delivery	none
no	NA	no					term delivery	none
no	NA	no			patient has pain over the elbow with restricted ROM at elbow with inability to hold heavy objects		term delivery	none

case 44		16	M	synostosis of radius and ulna	valgus deformity over right elbow	right	none	not done	normal vaginal delivery	no
case 45		20	F	post axial polydactyly	post axial polydactyly along the ulnar border of hand	left	bilateral feet- post axial polydactyly	not done	normal vaginal delivery	no
case 46	9 months	M		syndactyly with cleft hand	fused RARL digits with free thumb	right		not done	LSCS	no
case 47	7 months	M		bilateral radio-ulnar synostosis	Equinocavovarus deformity of right and left foot	right and left	none	not done	normal vaginal delivery	no
case 48		6	M	Sprengel deformity	winging of scapula with medialised hypoplastic scapula with muscle wasting over right scapular region	right	none	not done	normal vaginal delivery	no
case 49		1	F	congenital vertical talus	plano-valgus deformity of right foot	right	none	not done	normal vaginal delivery	no
case 50		1	F	bilateral equinocavovarous deformity	Equinocavovarus deformity of right and left foot	right and left	none	not done	normal vaginal delivery	no
case 51		1	M	bilateral metatarsus adductus	in toeing of bilateral feet	right and left	none	not done	normal vaginal delivery	no
case 52		3	M	bilateral equinocavovarous deformity	Equinocavovarus deformity of right and left foot	right and left	none	not done	normal vaginal delivery	no
case 53		4	M	left equinocavovarous deformity	plano-valgus deformity of left foot	left	none	not done	normal vaginal delivery	no
case 54		5	M	syndromic child with bilateral mirror foot on both sides with supra-numerary toes. B/L tibia h	bilateral mirror foot on both sides with supra-numerary toes. B/L tibia hemimelia with syndactyly of bilateral hands	right and left	none	not done	normal vaginal delivery	no
case 55	9 months	M		pre-axial polydactyly in bilateral hands	extra digits over the radial aspect of bilateral thumbs	right and left	scoliosis with PDA	yes- PDA- 1 mm, IVS with VWA	normal vaginal delivery	no
case 56		7	M	bilateral radio-ulnar synostosis	deformity of bilateral upper limb with restricted supination and pronation	right and left	intellectual disability	not done	normal vaginal delivery	no
case 57		3	F	radio-ulnar synostosis	deformity of left upper limb with restricted supination and pronation	left	none	not done	normal vaginal delivery	no
case 58		2	F	radial club hand with hypoplastic thumb with right microtia	deformity of right radial, hand and right ear	right	right ear-microtia	yes 2D ECHO & USG-WA- WNL	Normal vaginal delivery	no
case 59	10 months	F		bilateral equinocavovarous deformity	Equinocavovarus deformity of right and left feet	right and left	none	not done	normal vaginal delivery	no
case 60	1 day	F		bilateral equinocavovarous deformity with congenital hydrocephalus	Equinocavovarus deformity of right and left foot with macrocomia	right and left	hydrocephalus (dilatation of third ventric	Not done	LSCS (35 WEEKS +1 DAY)	NO
case 61	2 months	M		congenital femoral deficiency with ? Congenital fibular hemimelia with ? Left DDH with duplic	short left lower limb with extra fused great toe	left	sacral facies with bilateral undescended	2D echo- perimembranous VSD,	LSCS	no
case 62		2	M	case of trimax syndrome with persistent dissection of bilateral knees	deformity over bilateral knee with hyperextension at knees	right and left	becak rounded tips of fingers of bilateral	2d echo and USG WA- WNL	normal vaginal delivery	NO
case 63		10	M	post-axial Polydactyly bilateral hands	extra digits over the ulnar aspect of bilateral little fingers	right and left	none	not done	normal vaginal delivery	no
case 64	1 month	F		post axial polydactyly bilateral gaiters and feet with calcaneo-plano valgus left foot and cavo-v	extra digits bilateral and feet with equinocavovarous deformity on right side and calcaneo-plano valgus on left	right and left	none	Not done	normal vaginal delivery	no
case 65	1 month	F		right radial club hand with thumb in plam left side	deformity of right upper limb with absent thumb	right	multiple pustular rash over limbs and so	ocular examination-WNL, hearing	LSCS	NO
case 66		11	M	arthrogryposis multiplex congenita with multiple joint contractures	hypocalcaneus over left knee with flexion contractures with spina deformity	left	none	Not done	normal vaginal delivery	no
case 67	3 months	M		right congenital talipes equinocavovarous	equinocavovarus deformity over right foot	right	none	Not done	normal vaginal delivery	no
case 68		2	M	right congenital talipes equinocavovarous	equinocavovarus deformity over right foot	right	none	not done	normal vaginal delivery	no
case 69	6 months	M		right congenital talipes equinocavovarous	equinocavovarus deformity over right foot	right	none	Not done	normal vaginal delivery	no
case 70	2 months	F		left equinocavovarous deformity	equinocavovarus deformity over left foot	left	none	Not done	normal vaginal delivery	no
case 71		1	M	right congenital talipes equinocavovarous	equinocavovarus deformity over right foot	right	none	not done	normal vaginal delivery	no
case 72	8 weeks	M		bilateral congenital talipes equinocavovarous	equinocavovarus deformity of right and left feet	right and left	none	Not done	LSCS	no
case 73	6 weeks	M		right congenital talipes equinocavovarous	equinocavovarus deformity over right foot	right	none	Not done	normal vaginal delivery	no
case 74		1	M	bilateral congenital talipes equinocavovarous	equinocavovarus deformity of right and left feet	right and left	none	Not done	normal vaginal delivery	no
case 75		3	M	bilateral calcaneovalgus deformity	bilateral congenital vertical talus	right and left	none	Not done	normal vaginal delivery	no
case 76		2	M	mesodactyly of 3rd toe left foot	mesodactyly of 3rd toe of left foot	left	none	Not done	normal vaginal delivery	no
case 77		10	F	radiresidary 4th toe of right foot	tersal and radiresidary 4th toe right foot	right	none	not done	normal vaginal delivery	no
case 78		1	F	pterygium syndrome with deformity over right knee and foot	fixed flexion deformity of right knee with webbing of toes of right foot	right	none	2d echo and USG WA- WNL	normal vaginal delivery	no
case 79		1	M	radimesiary great toe left foot	pre-axial radimesiary toe	left	none	2d echo and USG WA- WNL	normal vaginal delivery	no
case 80		8	M	radio-ulnar synostosis-left side	deformity over left forearm with difficulty in pronation as supination	left	none	Not done	normal vaginal delivery	no
case 81		36	F	pre-axial polydactyly right hand	extra digit over the radial aspect of thumb	right	none	Not done	normal vaginal delivery	no
case 82	4 months	M		radial club hand with hypoplastic thumb left side	deformity of left forearm with hypoplastic thumb	left	none	Not done	normal vaginal delivery	no
case 83	7 months	M		bilateral congenital talipes equinocavovarous	equinocavovarous deformity of right and left feet	right and left	none	Not done	normal vaginal delivery	no
case 84		5	M	bilateral congenital talipes equinocavovarous with bilateral clasp thumb	equinocavovarous deformity of right and left foot	right and left	none	Not done	normal vaginal delivery	no
case 85	5 months	M		equinocavovarous deformity of left foot with flexion contractures of left knee	equinocavovarous deformity of left foot	left	none	Not done	normal vaginal delivery	no
case 86	4 months	F		equinocavovarous deformity of left foot	equinocavovarous deformity of left foot	left	none	Not done	normal vaginal delivery	no
case 87		4	F	equinocavovarous deformity of left foot	equinocavovarous deformity of left foot	left	none	Not done	normal vaginal delivery	no

NA	no			patient has pain over the elbow with restricted ROM at elbow with inability to hold heavy objects		term delivery	none	
NA	yes	surgical	excision of left hand extra digit	the extra finger would cause difficulty in doing household chores and f Ayushman Bharat		do not recollect	none	
NA	no			patient is able to hand pick the objects but parents were concerned about associated birth defects if any		term delivery	none	
NA	yes	non surgical	serial casting followed by application of foot abduction brace	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
NA	yes	non surgical	shoulder and back ROME and MSE	cosmesis and less power in right upper limb	APL-15000 INR	term delivery	none	
NA	yes	non surgical	reverse ponseti casting	abnormal positioning of right foot causing imbalance while walking	APL-15000 INR	term delivery	none	
NA	yes	non surgical	serial casting-ponseti technique- every week	abnormal positioning of right and left feet	APL-15000 INR	term delivery	none	
NA	yes	non surgical	stretching exercises	in toeing of bilateral feet		term delivery	none	
NA	yes	non surgical	serial casting followed by application of foot abduction brace	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
NA	yes	non surgical	serial casting followed by application of foot abduction brace	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
NA	yes	surgical	excision of 2 pre axial digits of both feet followed by metatarsal osteotomy	stabilization with k wire on right side, re aligning of third metatarsal as	Ayushman Bharat	term delivery	none	
NA	no		on follow up	parents were worried about the associated anomalies and wanted assurance		term delivery	none	
NA	no					term delivery	none	
NA	no					term delivery	none	
NA	yes	surgical	soft tissue release(radial and ulnar carpal ligament release) with PCR & E	restricted use of right hand and forearm + cosmesis	Ayushman Bharat	term delivery	none	
NA	yes	non surgical t/b surgical	ponseti casting. 0 times followed by tenotomy	cosmesis and future difficulties in walking		term delivery	none	
NA	no	advised casting at 3 weeks of age				9 months 29 days (term delivery	history of gestational diabetes and hypothyroidism	
NA	not till now					term delivery	none	
NA	Yes	surgical	patient underwent surgery for bilateral dislocated hips in 2023 and multipl	inability to use bilateral lower limbs	APL-15000 INR	term delivery	none	
patieral side has s no						term delivery	none	
no	yes	non-surgical for bilateral feet deformi serial casting		cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	patient admitted in SMTI medicine					pre term delivery	congenital rubella syndrome- TORCH infection	
no	yes	surgical	patient underwent multiple soft tissue release around left knee	difficulty in walking	ayushman	term delivery	none	
no	yes	non surgical	serial casting , ponseti technique every week	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	yes	non surgical	serial casting- ponseti technique-every week	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	yes	non surgical	serial casting-ponseti technique- every week	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	yes	non surgical	serial casting-ponseti technique- every week	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	yes	non surgical	serial casting- ponseti technique-every week	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	yes	non surgical	serial casting-ponseti technique -every week	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	yes	non surgical	serial casting- ponseti technique every week	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	no					term delivery	none	
no	no					term delivery	none	
no	no					term delivery	none	
no	yes	surgical	Ilizarov fixator application for gradual deformity correction	cosmesis and future difficulties in walking	Ayushman	term delivery	none	
no	yes	surgical	excision of supernumerary toe and fixation with k-wire	cosmesis	Ayushman	term delivery	none	
no	no					term delivery	none	
no	no					term delivery	none	
no	not till now					term delivery	none	
no	yes	non surgical	serial casting ponseti technique- total 3 casting done till now	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	yes	non surgical	serial casting to stretch out soft tissue- to be planned for a-la-carte soft tis	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	yes	non surgical	serial casting ponseti technique- 7 times till now	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	yes	non surgical	serial casting- ponseti technique	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	
no	yes	non surgical	serial casting-ponseti technique	cosmesis and future difficulties in walking	APL-15000 INR	term delivery	none	

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